Disabling Pregnancy:
Revealing how prenatal testing messages shape reproductive choices

Anjali Truitt

A dissertation

submitted in partial fulfillment of the

requirements for the degree of

Doctor of Philosophy

University of Washington

2015

Reading Committee:

Sara L. Goering, Chair

Wylie Burke

Janelle S. Taylor

Program Authorized to Offer Degree:

Public Health Genetics

University of Washington

Abstract

Disabling Pregnancy:
Revealing how prenatal testing messages shape reproductive choices

Anjali Truitt

Chair of the Supervisory Committee:
Associate Professor Sara L. Goering, Ph.D.
Department of Philosophy

Amniocentesis or chorionic villus sampling to diagnosis Down syndrome has long been an exemplar for prenatal genetic testing. With nearly 50 years of clinical application, the public has some awareness, knowledge, and expectations about such tests and Down syndrome. Yet, current evidence suggests that accepting or refusing testing is complex; the rationales for and narratives about doing so are nuanced. This scholarship highlights that patient expectations about commonly accepted tests are not necessarily always well-informed. While some enter this process as informed consumers seeking information, many accept testing with the hopes of reassurance about a healthy baby, and others accept a routine offer from their clinician. I qualitatively explore policy statements, health education, radio, newspapers, YouTube videos, and epidemiological data about Down syndrome birth prevalence to illuminate the interrelated,

normative expectations about parental responsibilities, testing utility, and reproductive choices. Across all data types, repetitive medical language about Down syndrome and limited discussion about testing purpose, risks, and benefits may offer inadequate or misinformation about such testing, making it difficult for risk-benefit assessments. Sources often frame prenatal testing as offering valuable, personal knowledge. They also suggest that prenatal testing provides early detection and disability prevention by means of pregnancy termination. In order to empower patients with the information and support needed to make the best reproductive choices for themselves and their families, we need: to ensure accurate and up-to-date information about Down syndrome and other prenatally diagnosable conditions is readily available; to include diverse perspectives in related health education and communication; and to direct attention to access issues that may constrain these choices.

TABLE OF CONTENTS

LIST OF FIGURES

LIST OF TABLES

LIST OF APPENDICES

ACKNOWLEDGEMENTS

I would like to acknowledge Dr. Sara Goering for her mentorship throughout this journey. Her thoughtfulness, inspiration, and friendship helped me to better consider alternatives and decide how to address challenges. Her insights and constructive feedback improved the writing quality and clarity immensely.

I would also like to acknowledge my other committee members. Dr. Wylie Burke for her guidance and direction towards concrete, real world examples. Her attentiveness and insights helped build my own confidence as a scholar. Dr. Janelle Taylor for pushing me to think more deeply about contextual issues and to challenge my own assumptions. More importantly, I appreciate her reminding me to celebrate even small victories. Dr. Sharan Brown for allowing me to stop by her office and check-in. Her willingness to chat about this, that, and everything, helped me to recharge.

Much appreciation to my second coder, Mr. Michael Nguyen. Without his tireless efforts, the quality and accuracy of this work would be much compromised.

I would like to thank the Institute for Public Health Genetics and the Disability Studies Program for their support on many levels throughout this process. Students, faculty, and staff have contributed to this project in many meaningful ways. I would like to especially thank Mr. Dennis Lang for his mentorship. His continued interest and willingness to support my work regardless of time or location has been invaluable. Dr. Joanne Woiak and Ms. Beth Secor for their friendship and continued support. To my colleagues in Public Health Genetics, particularly

Ms. Mercy Laurino who has been an ongoing motivation and inspiration to complete this project, I want to express my deepest gratitude.

Much love and thanks for all the support from my family and friends, especially Mrs. Priscilla Dawson and Ms. Shanda Truitt.

Without the support I received from these colleagues, friends, and family, this work would have been insurmountable. Their contributions made this possible.

This work is funded in part by the Harlan Hahn Endowment Fund awarded by the University of Washington Disability Studies Program. I appreciate its financial support for this project. The content is solely the responsibility of the author and does not necessarily represent the views of the funding source.

CHAPTER 1 INTRODUCTION

The best-selling pregnancy book What to Expect When You're Expecting is "America's Pregnancy Bible", according to the cover (Murkoff & Mazel, 2008). In its fourth edition, with over 15 million copies sold, it is difficult to deny its popularity. The book provides answers to hundreds of pregnancy-related questions from expectant parents, and as obstetrician-gynecologist Dr. Lockwood articulates, "[…] is like having a personal obstetrician to guide you through that adventure" (Murkoff & Mazel, 2008, p. xx). As the title indicates, the explicit point is to disclose to prospective parents what happens during pregnancy, so that they are prepared with information and knowledge about what to expect. More implicitly, however, it explains what prospective parents should and by default should not do, and what expectations and assumptions about pregnancy they should and should not have from preconception to postpartum. To me, this book provides just one example of a growing number of instances, where health communication and education is being offered outside patient-provider communication and beyond the clinic. While other sources may be less explicit, they still inform their audience about pregnancy and prenatal care, creating particular understandings about parental responsibility, utility of specific healthcare practices, and what constitutes health-promoting behaviors.

While What to Expect When You're Expecting offers a week-by-week pregnancy guide, my work takes on a more narrowly defined part of this process for most expectant parents in the United States: the offer of screening for chromosomal abnormalities, the screening results, a choice about diagnostic testing, the diagnostic results, and subsequent pregnancy management decisions. Because prenatal testing for Down syndrome has been around for nearly 50 years (Nadler, 1968), the public has at least to some extent awareness, knowledge, and expectations

about both related tests and Down syndrome (Kalfoglou, Suthers, Scott, & Hudson, 2004). Yet, accepting or refusing testing is complex; the rationales for and narratives about doing so are nuanced (Browner, 2000; Browner & Press, 1996; Browner & Press, 1995; Lippman, 1999; Markens, Browner, & Preloran, 2010; Markens, Browner, & Press, 1999; Press & Browner, 1994; Press & Browner, 1997; Rapp, 1993, 1996, 1998, 1999). This scholarship highlights that patient expectations about even commonly accepted tests are not necessarily always well-informed. While some enter this process as informed consumers seeking information about their pregnancy and developing fetus, many accept with the hopes of reassurance about a healthy baby, and others who simply accept a routine offer from their clinician. For some, the information received is that their developing child has a genetic condition and that they might consider terminating the pregnancy. Because of this diversity in patient knowledge, I am interested in how health messages about this process create and shape public understanding; particularly, I investigate how health messages convey interrelated, normative expectations about parental responsibilities, testing utility, and Down syndrome.

Such converging issues are by no means newly discussed in this work; however, I think several key changes to the context for which such offers and decisions are made make the work more pressing and interesting. First, the American College Of Obstetrics and Gynecologists (ACOG) has publicly communicated about a shift in clinical practice guidelines from testing only high-risk pregnancies for conditions like Down syndrome to offering testing to all pregnant women (American College of Obstetricians and Gynecologists, 2007a, 2007b, 2007c); this policy implies a series order from screening to diagnostic testing (Table 1), leaving the pathway largely up to patient-provider decision-making and specifying that the offer should be before 20

weeks gestation.[1] Second, broad shifts in public perception about utility have resulted in an expanded understanding about clinical utility that includes personal utility, particularly in the context of genetic information.[2] Third, developments in our understanding about biophysiology and its implications has resulted in improved risk assessment for Down syndrome prenatally (i.e., high-resolution ultrasonography, cell-free fetal DNA testing).[3] Fourth, changes in healthcare delivery have resulted in a multitude of diverse health messages about this process; specifically, a move from clinic-based patient education for prospective parents considering prenatal testing for Down syndrome towards computer-assisted educational strategies and broader online health communication and education on these topics.[4] Lastly, societal changes at a population-level about timing of child-bearing may influence the number of prospective parents accepting such testing and receiving a prenatal diagnosis of Down syndrome.[5] As developments in prenatal screening methods improve risk assessment for Down syndrome and broaden the population who might be offered clinical testing, an increasing number of patients may seek information about why they should accept or refuse testing from an array of sources in their decision-making process. At the same time, the public narrative is at a turning point, where shifts in knowledge and expectations are evident. To me, understanding how health messages might shape expectations is essential to ensuring that patients are well-informed and empowered to make value-consistent decisions. Ultimately, this work illuminates at this point of transitions how

[1] Chapter 2 provides a more complete description and analysis of this policy.
[2] Later in this Chapter, I layout some general constructs about clinical utility. Chapter 4 delves further into how the radio creates this construct and public reaction to this.
[3] Chapter 5 discusses cell-free fetal DNA testing more specifically and how newspapers portrayed this "new" methodology in contrast to existing ones.
[4] Chapter 3 analyzes clinic-based brochures and online resources, as well as broader public health education materials available online. Chapter 6 analyzes YouTube videos to highlight somewhat different strategies and language are used in this community, video-sharing resource.
[5] Chapter 7 describes the epidemiological evidence underpinning this argument.

health messages – in policy, newspapers, radio, health education, and YouTube videos – shape public knowledge and expectations, implicitly and explicitly, about prenatal screening and diagnostic testing for Down syndrome in the United States.

Table 1. Overview of prenatal testing methods for Down syndrome detection

Method	Description	Components examined	Testing timeframe (gestational age, weeks)	Detection rate (in ascending order)	False-positive rate
Screening options					
Maternal Age	Age of mother	Age-related risk	N/A	26% (Wald, Huttly, & Hackshaw, 2003) 30% (Nicolaides, 2011) 51% (Wald et al., 2003)	5% (Wald et al., 2003) 5% (Nicolaides, 2011) 14% (Wald et al., 2003)
Nuchal translucency scan (NT)	Ultrasound scan of fetus identifying subcutaneous fluid collection behind the fetal neck	Fluid translucency behind fetal neck and assessment of nasal bone	12-13 (Bindra, Heath, Liao, Spencer, & Nicolaides, 2002) 11-13 (Nicolaides, 2011)	70% (Nicolaides, 2011)	5% (Nicolaides, 2011)
First-trimester screening	Biochemical markers evaluated in maternal serum from blood test	Pregnancy-associated plasma protein-A (PAPP-A) and fetal βeta-human chorionic gonadotrophin (fβ-hCG)	11-14 (Bindra et al., 2002)	90% (Bindra et al., 2002) 91.2% (Leung *et al.*, 2009) 87% (11wks) 85% (12wks) 82% (13wks) (Leung et al., 2009)	5% (Bindra et al., 2002)

Method	Description	Components examined	Testing timeframe (gestational age, weeks)	Detection rate (in ascending order)	False-positive rate
First-trimester combined screening	Combination of first-trimester screening, NT and maternal age-related risk	PAPP-A, fβ-hCG, NT, maternal age	11-14 (Nicolaides, Spencer, Avgidou, Faiola, & Falcon, 2005) 10-14 (Spencer, Souter, Tul, Snijders, & Nicolaides, 1999)	70% (Spencer et al., 1999) 75% (Nicolaides et al., 2005) 80% (Nicolaides et al., 2005) 82% (Malone *et al.*, 2005) 85%* (Malone et al., 2005) 89% (Spencer et al., 1999) 90% (Nicolaides et al., 2005) 85-95% (Nicolaides, 2011)	1% (Spencer et al., 1999) 1% (Nicolaides et al., 2005) 2% (Nicolaides et al., 2005) 3.2% (Malone et al., 2005) 5.6%* (Malone et al., 2005) 5% (Spencer et al., 1999) 5% (Nicolaides et al., 2005) 5% (Nicolaides, 2011)

Method	Description	Components examined	Testing timeframe (gestational age, weeks)	Detection rate (in ascending order)	False-positive rate
Maternal serum screening: Quad screen	Biochemical markers evaluated in maternal serum from blood test	Alpha-Fetoprotein (AFP), total human chorionic gonadotrophin (hCG), unconjugated estriol (uE3), Inhibin A (INH)	15-18 (Leung et al., 2009)	65-70% (Nicolaides, 2011) 75% (Wald et al., 2003) 81% (Leung et al., 2009) 85%* (Malone et al., 2005) 81% (Wald et al., 2003)	5% (Nicolaides, 2011) 5% (Wald et al., 2003) 8.5%* (Malone et al., 2005) 7% (Wald et al., 2003)
Integrated panel	Biochemical markers evaluated in first and second trimester in maternal serum from blood test plus with nuchal translucency scan	AFP, total hCG, uE3, Inhibin A, NT, PAPP-A	14-22 (Wald *et al.*, 2009)	87% (Wald et al., 2009) 88% (Leung et al., 2009) 90-94% (Nicolaides, 2011) 96% (Integrated + first trimester screening) (Leung et al., 2009)	2.1% (Wald et al., 2009) 5% (Nicolaides, 2011)
Cell-free fetal DNA testing (cffDNA)	Circulating fetal DNA is collected from within maternal blood	Quantitative analysis of short fetal DNA fragments	11.6-25.7; mean 13.3-19.5 (Norton *et al.*, 2012)	98.6%* (Palomaki *et al.*, 2011) 100%* (Norton et al., 2012)	0.2%* (Palomaki et al., 2011) 0.03% (Norton et al., 2012)

Method	Description	Components examined	Testing timeframe (gestational age, weeks)	Detection rate (in ascending order)	False-positive rate
Diagnostic options					
Chorionic villus sampling (CVS)	Transabdominal or transcervical placental biopsy using ultrasound for guidance	Cytogenetic evaluation of placental tissue	10-13 (National Coalition for Health Professional Education in Genetics & National Society of Genetic Counselors, 2012)	99.99% (National Coalition for Health Professional Education in Genetics & National Society of Genetic Counselors, 2012)	< 1% (National Coalition for Health Professional Education in Genetics & National Society of Genetic Counselors, 2012)
Amniocentesis (amnio)	Withdrawal of amniotic fluid using ultrasound for guidance that is cytogenetically tested	Cytogenetic evaluation of fetal cells circulating within amniotic fluid	15-17 (American College of Obstetricians and Gynecologists, 2007d)	99.99% (National Coalition for Health Professional Education in Genetics & National Society of Genetic Counselors, 2012)	< 1% (National Coalition for Health Professional Education in Genetics & National Society of Genetic Counselors, 2012)

Asterisk (*) denotes data from pregnancies deemed high-risk, most often in relationship to maternal age at birth

Background Literature

In efforts to provide readers with at least a cursory background of the key concepts addressed in this work, I lay out some of the scholarly discussions about clinical utility, disability, and ethical considerations surrounding such testing. First, I describe some of the prevalent conceptualizations of clinical utility, particularly in relationship to genetic testing. Second, I lay out several models to understanding disability, because these socio-identity constructions of disability provide resistance to the pervasive medical understanding of disability and illuminate an alternative, often silenced means of understanding disability. Third, I discuss some of the disciplinary perspectives involved and highlight central values and ethical concerns about prenatal testing and its practice.

Clinical Utility

Genetic tests must have the ability to accurately and reliably identify the genotype of interest (i.e., analytic validity) and the capability to detect accurately and reliably the condition it is intended to identify (i.e., clinical validity) (Constantin, Faucett, & Lubin, 2005). Broadly, there is biomedical consensus with regards to the analytic validity and clinical validity of prenatal genetic testing for Down syndrome; that is, available genetic tests accurately and reliably identify trisomy 21 and this genetic trait is associated with clinically identifiable features that are characteristic of Down syndrome. Prenatal genetic testing for Down syndrome becomes more complicated when considering clinical utility, in that there is limited consensus about the meaning of clinical utility and even less consensus about exactly how prenatal testing for Down syndrome fits these understandings. Traditionally, research applications move to clinical care when the application can improve patient outcomes. From a clinical perspective, there are three common scenarios in which an application is considered to have clinical utility: first, when the application can prevent or cure a condition; second, when the application can reduce morbidity;

third, when the application can guide treatments. These clinically actionable outcomes make the move from research to clinical care justifiable (Zimmern & Kroese, 2007). At present, there is no prenatal therapy available to treat or cure Down syndrome. Strictly speaking, these scenarios do not apply to prenatal testing for Down syndrome. Despite this, such testing is routinely offered and widely accepted.

Particularly with regards to genetic testing, some have discussed the need for considerations about personal utility (Khoury *et al.*, 2009). While clinical utility of a genetic test requires an effective intervention or other clinically actionable outcome, personal utility suggests that a genetic test should minimally yield some type of useful genetic or other health information. There are several reasons we may consider a test to have personal utility: it allows an individual to prepare themselves and/or their families, to arrange personal affairs, to guide reproductive decisions, etc. Personal utility is often discussed in relationship to the REVEAL study, an evaluation of genetic communication to people at-risk for Alzheimer disease (Chao *et al.*, 2008; Green *et al.*, 2009; LaRusse *et al.*, 2005; Roberts *et al.*, 2004; Zick *et al.*, 2005). These studies show that some participants found this risk information personally useful, even though there is no effective intervention available to reduce risk. They also revealed that participants did not experience any major adverse psychological effects from knowing their risk status, even in instances where participants were identified as having high-risk. Worth noting, the participants were adult children who had a parent with Alzheimer's disease, so they were already aware of their family history with the condition of interest.

In light of conversations about personal utility, even among health professionals, there has been expanding understanding about the meaning of clinical utility. A more general definition of clinical utility might call for a balance of risks and benefits (e.g. psychological harm

and therapeutic benefit) from both positive and negative results; that is, a test has clinical utility if either the provider or the patient does something more than worry based on test results (Task Force on Genetic Testing, Holtzman, & Watson, 1998). While the traditional understanding of clinical utility focuses on clinically actionable outcomes, this definition includes personal outcomes as equally valuable. This understanding later expanded to include short and long-term outcomes that included psychological, social, family and cost-benefit assessments (Secretary's Advisory Committee on Genetic Testing, 2000). Clinical utility has also been discussed as the likelihood of a test to result in improved health outcomes, highlighting the need to consider the broader context of a test (e.g. is genetic testing the only mechanism to identify the trait; are effective intervention available; what ethical, legal, or social consequences might testing have; is pre-and post-tests counseling available; is an equitable access to testing; what are the financial costs) (Burke *et al.*, 2002; Zimmern & Kroese, 2007). Based upon this understanding, even a test that fits the traditional definition may not actually have clinical utility in practice, due to its broader impact and implications. Still others have described clinical utility from a population perspective, so-called public health utility, to mean an absolute or risk reduction in burden of disease (Khoury, Jones, & Grosse, 2006).

Prenatal genetic testing for Down syndrome has already departed from traditional considerations about clinical utility and moved towards personal utility. The primary goal of prenatal genetic tests centers upon the ability to provide information to prospective parents, and in this instance, the information itself is framed as an endpoint, rather than viewing a clinical action as an endpoint (Press, Wilfond, Murray, & Burke, 2011). In fact, there are three main reasons why prospective parents choose prenatal genetic diagnosis: 1) to terminate a fetus identified with Down syndrome; 2) to gain advance notice about Down syndrome in order to

prepare for raising a child with the condition; or 3) to initiate adoption services (Skotko, 2009). According to one systematic review, about 90% of parents decide to terminate a fetus identified with Down syndrome (Mansfield, Hopfer, & Marteau, 1999). From a public health perspective, prenatal screening and testing for Down syndrome has led to an overall reduction in the US incidence of the condition (Egan *et al.*, 2004). In fact, worldwide birthing trends indicate that women are delaying childbirth and thus advancing maternal age. Because of the positive association between advancing maternal age and Down syndrome, we would expect more infants born with Down syndrome now compared with 10 years ago, but instead, we have seen an overall decrease in incidence rates (Skotko, 2009). Thus, prenatal genetic testing may yield both personal utility and public health utility.

One of the central issues of my work is uncovering how utility is used in health messages about prenatal testing, in order to identify publicly expressed and personally held values about utility. I want to expose how within the public sphere these constructs are produced and recreated outside the constraints of scholarly discourse. While clinical utility and personal utility are both useful organizing frameworks by which to classify how a particular perspective conveys the utility of a particular emerging technology or diagnostic test, these specific words may be absent from health messages. Alternatively, health messages may conflate clinical and personal utility. In doing so, patients may accept testing thinking it is of clinical utility, even though the offer may be intended to enhance personal utility. In this manner, identifying ways to disentangle clinical and personal utility may have particular relevance to improving informed consent of patients offered such tests. As such, I investigate the extent to which they are used, but more importantly, how clinical utility, personal utility, and even more broadly, the balance of the risks and benefits of such testing is articulated.

DISABILITY

In the US and Europe, antiwar and civil rights movements of the 1960's helped to organize activism and advocacy around disability-related issues. In other countries, leftist politics and national liberation further advanced what would become the disability rights movement (DRM). In this regard, the DRM was a social and political movement that aimed to promote inclusion, empowerment, and full participation of people with disabilities in their communities.

In the 1970s, the Union of Physically Impaired against Segregation, a prominent disability rights group in the United Kingdom, made an explicit distinction between the notion of impairment and that of disability. Building from this distinction, Oliver coined the terms individual and social models of disability (Oliver, 1996). The individual model or medical model suggests that disability is a personal problem or personal tragedy that necessitates individual treatment through medical intervention; from this framework, health care professionals are given domain over bio-physiological human differences. Oliver termed these bio-physiological human differences impairments. In direct contrast, the social model suggests that disability is a form of social oppression; disability is a social problem. As such, disability requires social action, self-help, and collective responsibility (Oliver, 1990). Similar to gender studies scholarship, impairment was equated with the biological, like sex; disability was equated with the social, like gender. However, Oliver went even further, because he suggested that disability is itself the experience of oppression (Oliver, 1990).

Based on this individual model of disability, functional limitations cause problems associated with disability. In contrast, the social model of disability attributes problems associated with disability to society, rather than individuals with disabilities; by failing to provide appropriate accommodations and services, society creates problems for people with

disabilities (Oliver, 1996). In this regard, society's oppression is the cause of disability, not an individual's physical, mental, or behavioral differences, and this understanding of disability is based upon personal experience of living with a disabling condition rather than perspectives from medical authorities.

The individual model of disability dominates much of the public narrative about disability. In fact, Disability Studies scholars in media studies highlight two important ways in which the individual model of disability surfaces: first, depictions of people with disabilities as children and second, representations of people with disabilities as pitiable (Haller, 2010; Riley, 2005; Shapiro, 1993). I think that this area of scholarship is particularly relevant to my analysis, because it highlights that the individual model has multiple, yet distinct manifestations in the media; furthermore, these manifestations show how media perpetuates stereotypes about disability that may contribute towards discriminatory social attitudes.

In contrast, consider a specific example of the social model; deafness (versus hearing) within the Deaf community highlights how when a particular aspect of physical functioning is viewed as binary, broader understandings of health may be overlooked. In her book Everyone Here Spoke Sign Language, Groce (1985) describes the timeframe in Martha's Vineyard where deafness was so common that everyone – hearing and deaf alike – used signed language; when everyone used sign, deafness was not disabling. Here, individuals with deafness had control and autonomy of their own decisions and were not made to be dependent. In fact, they were often more educated than their peers without deafness. This example shows how dominant social attitudes may impact our understanding of disability.

While these models of understanding disability are contested and expanded upon within Disability Studies literature, these guiding frameworks illustrate possible framings of Down

syndrome in health messages about prenatal testing. A medical model framing might focus on Down syndrome as a genetic trait and emphasize the clinical features or "problems" associated with the condition, including mental retardation, characteristic facial features, or heart defects. Given the absence of cure or treatment for Down syndrome, the individual and their family must "overcome" these problems, in order to have the valued "normal" life. In contrast, a social model framing might suggest that trisomy 21 is one of many possible human genetic variations, contributing to uniqueness and identity, like race or ethnicity might. The key distinction is that the social model focuses attention on how society disables people with Down syndrome, rather than the physical and mental characteristics of the individual. Consider school achievement. The medical model suggests that Down syndrome is associated with lower IQ, and this is the primary cause for underachievement. In contrast, the social model suggests that discriminatory attitudes (e.g., assumptions that people with Down syndrome cannot succeed in school or will perform poorly) and exclusionary practices (e.g., special education classes where people with Down syndrome receive segregated and thereby unequal course instruction) are more significant to the cause of underachievement than lower IQ itself. In other words, the point of the social model is to redirect attention towards how society and social structures negatively impact the lives of people with disabilities as much or perhaps more so than the underlying physical or mental traits.

Ethical and Social Issues

Broadly, the central issue with regards to prenatal testing relates to patient autonomy. Scholars raise a variety of concerns addressing this, but there are nuances about their arguments worth discussing. Particularly across genetic counseling, public health and bioethics, professional duties and disciplinary values about what matters most focus their concerns in particular ways. While I recognize that I am in many ways creating caricatures of these

disciplinary perspectives, I hope to highlight some useful distinctions that I believe create tensions and confusions in the public sphere. Understanding these academic conversations, which are often used to influence policy and public opinion, has important implications for public understanding about prenatal genetic testing, including the understanding about potential risks and benefits.

Medical Genetics

As Davis (2010) articulates, the field of genetic counseling epitomizes respect for patient autonomy more than other healthcare professions. Davis explains several factors that she thinks contribute to this focus. First, she attributes this focus to the fact that the medical genetics community understands its historical connection to eugenics. Second, the medical genetics community recognizes that the public may perceive new reproductive technologies as promoting commodification (i.e., parents might pick certain traits about their potential child that they want and deem other traits undesirable and use of in vitro fertilization to select embryos that meet these criteria or selectively terminate pregnancies that do not). Third, the medical genetics community is aware of growing concern about so-called laissez-faire eugenics, which suggests that individual, consumer-driven reproductive choices taken collectively may have the same impact and intolerance towards differences as the government coercion of Nazi era eugenics. Within genetic counseling, the focus on patient autonomy has led to the development of two central tenets: value-neutrality and nondirective counseling. Taken together, the value of a decision is solely based upon what is right for that particular patient and counselors should not try to influence a decision (Davis, 2010; Madeo, Biesecker, Brasington, Erby, & Peters, 2011); however, these values are not held to as strongly by other professionals involved with the delivery of health education and communication about prenatal testing.

Many authors have expressed concerns about the context of the offer of prenatal genetic testing, albeit there are subtleties with regards to what about the context is concerning. While a multidisciplinary group of scholars has expressed these concerns, most focus on the patient-provider relationship. OB/GYNs are increasingly and more consistently involved with the offer of prenatal genetic testing. This shift in who delivers the offer of testing is an expressed concern, because by and large, OB/GYNs do not share as strong a value on nondirectiveness as those with training in genetic counseling (Marteau, Drake, & Bobrow, 1994; Marteau, Plenicar, & Kidd, 1993). As such, OBs may be more accustom to offering a recommended procedure and expecting patient compliance −possibly making the offer seem like a recommendation− than genetic counselors who are trained to hold value neutrality about offered tests and nondirective counseling. Even when genetic counselors are involved, some contend that patients do not make substantially different decisions from before to after a genetic counseling session, suggesting that people have set preferences and do not change their mind with genetic counseling; instead, genetic counseling is really about addressing the psychosocial needs of patients, including the need for support and reassurance about their decisions (Biesecker & Hamby, 2000; Madeo et al., 2011).

Several authors have expressed explicit concerns with healthcare providers' knowledge about disability. Like the general population, public representations of disability bombard healthcare providers in ways that can misinform and misinterpret the experience of living with a disability. According to Asch (2003), there are three dominant assumptions that fuel acceptance of prenatal genetic testing, diagnosis, and selective termination: first, the disadvantages that people with disabilities experience stemmed from their impairments, rather than from discriminatory attitudes and social structures that prevent people with disabilities from full

participation in the community; second, the emphasis on the range of opportunity as opposed to the possibility of meaningful choices and rewarding outcomes within any range; third, confusion about how having a particular trait is good means that lacking a particular trait must be bad. Yet, people with intellectual/developmental disabilities, including Down syndrome, report having fairly independent and rewarding lives (Alderson, 2001). While healthcare providers admitted limited exposure to disability or genetics, they expressed comfort when talking about these genetics (59%) and disability (75%). They also expressed that disability causes suffering to the individual and their family (51% and 64%, respectively) (Ormond, Gill, Semik, & Kirschner, 2003). In fact, recent research on health care providers' attitudes suggests that healthcare providers view prenatal diagnosis and selective termination more favorably than their patients (Klein, 2011). By portraying disability as causing suffering and promoting selective termination, prospective parents may feel a responsibility to accept both prenatal testing and selective termination.

In order for patients to make an informed, autonomous decision about genetic testing, they must have an understanding of the purpose of the test, the potential risks and benefits of both choosing to test or not to test (or alternative options, if they exist), and the freedom to choose any option(s). Typically, the patient should undergo a process of informed consent for both prenatal screening and subsequent diagnostic tests. Pilnick illuminates how the presentation format may unintentionally lead patients to accept screening. Specifically, she highlights that often within conversations with health care professionals about screening, the offer is not straightforward; patients do not always understand that they are being offered screening or not in comparison to "new" versus the "old" screening methods (Pilnick, 2004). In other words, some patients may not understand when healthcare providers are giving them an opportunity to make a

decision about whether to accept or reject prenatal testing. Especially for people with lower numeracy literacy, using frequency (i.e. comparing one out of 724 fetuses of women your age to one out of 181 fetuses of women your age), rather than 1-in-N (i.e., comparing 1:724 to 1:181) or visual formats (i.e., comparing a picture with a white circle amidst 723 black circles to a picture of one white circle amidst 180 black ones) proved to increase understanding about risk assessment of having a fetus identified with Down syndrome (Miron-Shatz, Hanoch, Graef, & Sagi, 2009). This suggests that some patients are overestimating the possibility of having a fetus with Down syndrome, which may further promote testing. In fact, a recent study shows when pregnant women could accurately identify the risks associated with amniocentesis, they were much less likely to accept testing (Kuppermann *et al.*, 2015). This suggests that many pregnant women may be inadequately informed before accepting such testing and that decisions might change with better information – not about disability, but simply about the testing methodologies available.

Additionally, some women are accepting testing for the reassurance that their developing baby is healthy, but do not fully understand that a "positive" result may lead to conversations about pregnancy termination (Press & Browner, 1997). Furthermore, not all patients read health education materials provided about the testing, and even when they do, patients do not always understand what conditions are being tested for or that there is no intervention available to treat them (Rothman, 1994). Also, worth noting, is the fact that within pediatric and prenatal genetic counseling, healthcare providers give dramatically different descriptions about the same conditions. In the prenatal context, counseling sessions focused upon medical differences; whereas, in pediatric counseling sessions, counselors focused on similarities between children with genetic conditions and their nondisabled peers (Lippman & Wilfond, 1992). These authors

highlight the concern with the provision of information for prospective parents offered this testing. This issue is central to my dissertation. While I have situated this issue as a concern for genetic counseling, I think that public health professionals will need to be increasingly aware of these concerns, as well.

PUBLIC HEALTH

In From Chance to Choice, the authors propose a public health model for genetic intervention. In contrast to a personal choice model, public health emphasizes the creation of benefits and the avoidance of harms to groups, which may focus its attention away from issues of discrimination and justice. Because of this, public health often takes a cost-benefit or cost-effectiveness approach towards genetic interventions. The field as a whole relies dominantly upon a consequentialist ethical framework (Buchanan, Brock, Daniels, & Wikler, 2000). In addition to the common history of eugenics, US public health has had its own negative history with the implementation of genetic interventions for carrier screening of sickle cell anemia (Markel, 1997). Because of public health's success with reducing infectious disease, prevention of disease transmission has become a central tenet of the field ("Ten Great Public Health Achievements--United States, 1900-1999," 1999). However, this central ideology is problematic when applied to genetic conditions. For example, strategies like mandatory vaccinations and quarantine explicitly infringe upon individual choice (Buchanan et al., 2000). Within the field of public health, restrictions on individual freedoms for the greater good are considered both appropriate and justifiable tools to prevent disease.

Safeguarding women's health rights is a dominant concern within public health. Global health is a growing field, and increasingly, we see interactions between US health policy and other countries. While in the United States, abortion is legal; in Brazil and other countries, it is not. Some have discussed whether it is morally permissible to offer prenatal genetic testing, if

termination services are unavailable (Ballantyne, Newson, Luna, & Ashcroft, 2009). While I think the premise of this argument is to ensure that all women have access to pregnancy termination services, such an argument implies that the clinical utility of prenatal genetic testing is dependent upon access to termination services. The argument also makes broader connections to the role of government. Because most public health interventions and programs generally involve government funding if not also infrastructural government support, some might argue that the engagement of public health with genetics raises concerns about eugenics. One of the expressed concerns of eugenics is statism. Buchanan et al. (2000) argued that government involvement itself does not make a genetic intervention or program eugenic; furthermore, they contend that removing the government may not prevent a program from having eugenic qualities. In fact, they suggest that Galton, the founder of the US eugenics movement, imagined a fully voluntary program. They argue that he hoped that the eugenics movement would compel individuals to make sacrifices for the public good (Buchanan et al., 2000).

Even when disregarding concerns about statism, public health interventions need to address moral considerations about paternalism (Childress *et al.*, 2002). Public health programs like newborn screening, or more broadly, seatbelt or helmet laws are paternalistic; yet, many consider such paternalism to be permissible in these instances, because of the perceived benefits to those affected (i.e., newborn screening all infants, so that children with particular genetic conditions are identified earlier, or requiring seatbelts or helmets, so that fewer people are injured in bicycle or automobile accidents). Instead of statism, some might argue that paternalism is embedded in eugenics movements, in that people in positions of authority restricted the freedoms of particular groups –specifically, people thought to be inferior or subordinate to those in authority– allegedly in the best interests of the subordinate group. For

example, laws that permitted medical superintendents of asylums and prisons to sterilize a patient or inmate, if it was deemed to improve her condition –physically, mentally, or morally (Stern, 2005). Like seatbelt or helmet laws, some might consider early 1920's American the paternalism embedded within sterilization laws morally permissible. However, non-English-speaking immigrants, Mexicans and African-Americans, and white women were disproportionately sterilized in comparison to English-speaking immigrants, white Americans, and white men, respectively (Stern, 2005). In this manner, paternalism was a guise for classism, racism, and sexism. Given histories of eugenics, some might question whether paternalistic genetic interventions and programs could ever be free of discrimination and thereby whether such paternalistic genetic interventions and programs should ever exist. Because the public understanding of eugenics is so negative and arguably lacks sophistication, eugenics is a real concern. However, part of the concern about eugenics is that people make comparisons to it without clearly defining what it is or means or how it is problematic.

Another of the dominant concerns public health professionals is resource allocation. If the incidence of Down syndrome is declining due to prenatal testing, some might argue that fewer resources are required to support children and families with Down syndrome. From the perspective of public health and public health utility, prenatal testing and selective termination together is arguably good; however, prenatal testing alone might be considered a waste of resources. The individual costs per patient may alone seem reasonable; however, from a public health perspective, it is unclear whether the expense of offering screening to all pregnant women is truly the best use of resources, in that lowering the disease burden by means of reducing incidence is the only mechanism to have a population level effect.

To my knowledge, prenatal testing is well-described ethically, but less so economically. In relationship to newborn screening programs, opportunity costs (i.e., the costs associated with directing resources in a particular direction at the expense of others) are well-described, so in order to better situate my discussion, I draw from this literature. When considering these costs, public health professionals explicitly take into consideration alternative scenarios that might yield greater benefits or distributing resources to another use might yield their benefits (Baily & Murray, 2008). Opportunity costs are greater given how many pregnant women are screened. Consider that the time (e.g., time spent informing and consenting patients), human resources (e.g., pre- and post-test genetic counseling), and cost of prenatal screening nearly all pregnant women and diagnostic testing most at-risk pregnancies is put towards alternative, health-promoting activities, like tobacco cessation. To the extent that tobacco use is a common issue, that cessation activities reduce maternal tobacco exposure, and that increasing the number of available cessation programs increases the number of women who actually stop using tobacco, we might imagine that such a switch in research allocation would improve population health more than prenatal testing. This is because pregnant women would decrease their long-term cancer risks and decrease their children's risk of a birth weight and asthma. In this manner, some might argue that in terms of resource allocation, prenatal testing may not be worth as much as tobacco cessation efforts.

When considering that recent research has shown promise with whole genome sequencing using cell free fetal DNA, like newborn screening, we may soon have the tools to test for many more genetic conditions prenatally than we currently do (Kitzman *et al.*, 2012). Using the framework of personal utility, some might consider any genetic testing appropriate if prospective parents deem the information important. In the prenatal context, we already test for

conditions that have limited or no evidence of benefit to the affected children (i.e., there is no prenatal care, treatment or intervention for fetuses affected with Down syndrome); resource allocation in this regard might become especially problematic. Taken together, it is foreseeable that we would test for a larger number of conditions with limited benefit. As discussed with regard to newborn screening, this may in fact cause harm, for the cost of testing and provisions of genetic counseling will likely squander scarce resources (Botkin *et al.*, 2006). Again, offering prenatal genetic screening that identifies more conditions may also mean that more patient-provider time will need to be allocated to discuss the implications of the testing, or perhaps, more time will need to be allocated to develop alternatives that address the range of conditions that might be identified prenatally, like interactive websites or patient-directed games. Regardless of the mechanisms, more time and efforts will be needed, as more people choose to test for a wider range of conditions.

Exploring beyond the clinical encounter, Saxton argues that the lack of social supports to assist parents raising a child with a disability make choosing to raise a child with Down syndrome increasingly difficult for prospective parents. In this manner, she contends that the social structures of our society lead parents to choose prenatal testing and selective termination. Without changing public provisions for children with disabilities and their families, we as a society, constrain parental choices about prenatal genetic testing and selective termination (Saxton, 2006). In contrast, newborn screening policy applies a specific financial charge to all newborns, in order to create a fund for medical services and treatment for infants identified with particular conditions. In doing so, this addresses some of Saxton's expressed concerns; therefore, public health professionals might consider a similar charge with regards to prenatal testing. Broadly, Saxton's discussion highlights the importance of the provision of services for

people with disabilities as an ethical consideration. While Saxton argues that a lack of social support constrains reproductive autonomy, Asch argues that unreflective use of prenatal testing may limit reproductive autonomy. Asch contends that identifying disabling traits may cause expectant parents to terminate "wanted" pregnancies. Asch suggests that the role of public health professionals is to prevent the inadequate social arrangements from which disability stems, like disability-based discrimination (Asch, 1999). The provision of services –namely, support services− for people with disabilities and their families is often overlooked, especially in conversations about prenatal genetic diagnosis.

BIOETHICS

Bioethicists may be less inclined towards a particular ethical framework or guiding principles than public health professionals (i.e., consequentialist) or those trained in genetic counseling (i.e., responsibilities to protect patient autonomy). In some ways, bioethicists have the freedom to consider the issue of prenatal genetic testing without consideration for these embedded professional duties. I recognize that bioethics is an interdisciplinary field, but I have purposefully selected ethical concerns non-healthcare providers raise. The considerations I discuss here extend what has already been stated by focusing on meaningful relationships and values in society. More broadly, I think that they reflect ethical considerations that are largely overlooked and add complexities to the overall conversation about prenatal genetic testing.

Bioethics often helps us to draw distinctions between two potentially similar actions and in doing so, helps to define and to provide clarity to a moral issue. Asch proposes the so-called expressivist argument, which has become the foundational Disability Rights critique of prenatal genetic testing. Asch argues that selective termination following prenatal genetic diagnosis is morally problematic; in part, because termination on the basis of a particular trait expresses discriminatory attitudes about the particular trait, but perhaps more importantly, about other

people who share the trait. Based upon her argument, selective termination is pregnancy termination on the basis of a particular trait, where this information is the primary reason prospective parents no longer want a previously wanted child. In this manner, she presents a distinction between abortion of an unwanted child and selective abortion of a wanted child who has an unwanted trait; thus, she creates space to maintain morally permissible cases of abortion. Her position finds that nondisabled people terminating fetuses identified with Down syndrome is as morally problematic as people with particular genetic conditions (e.g., congenital deafness or achondroplasia) terminating fetuses identified without the trait (Asch, 2000). Nelson (1998) argues against the expressivist argument, suggesting that the argument precludes an option for morally permissible abortion, given that any decision to abort is always a decision about a particular fetus. Asch (2000) refutes his claim, arguing that the particular trait is more than a characteristic of the fetus. The trait attaches particular social meaning, in that this very specific, limited information is sufficient to predict the child-rearing experience and to assess that a child with such a trait will not meet expectations; the trait alone is the basis for a decision to terminate (2000). Steinbock (2000) provides another counterargument to the expressivist argument. Specifically, she argues that there is no difference between selective termination and prevention. In order to illustrate this, Steinbock (2000) uses the example of spina bifida; she argues that selective termination of a fetus identified with spina bifida is no different from taking folic acid preconception, because in both instances, someone with a disability is not born. I find her argument particularly interesting, because of the potential connection to clinical utility (i.e., if selective termination is equated with prevention strategies, then prenatal genetic diagnosis might be considered clinically valid, even from a narrow definition of clinical utility). Throughout my

analysis, I will investigate to what extent this argument is represented in health message about such testing.

To me, some of the more interesting philosophical discussions about prenatal genetic testing address parent-child relationships, given that they illustrate some of the normative expectations about parental responsibilities and obligations to a child. In doing so, concerns about the parent-child relationships seem more relevant to public representations including family members of people with Down syndrome or people who choose to terminate following a prenatal diagnosis of Down syndrome. Unlike the expressivist argument, which focuses on selective termination, concerns about the parent-child relationship uncover interesting features about our values that I believe have broader applicability to what we consider the benefits and risks of genetic testing. For example, Davis (2010) argues that the developing fetus has a right to an open future. Her argument makes us consider whether people with particular genetic conditions should select to have a child with a similar trait. She contends that they should not, because selecting a particular trait might ultimately limit the child's range of options for future (Davis, 2010). Broadly, I think she helps us to think about the parent-child relationship, the power between the two, and parental duty to maintain options for a child. Based upon this, some might consider genetic tests for a wider range of pediatric onset conditions morally permissible or even morally required. Kittay & Kittay (2000) also discuss parental-child relationships; specifically, they suggest that prenatal genetic diagnosis might transform the family unit into a "club", where membership is based on having or not having a particular trait. This suggests parents have an obligation to be accepting, rather than exclusionary, and in the context of genetic testing, some might consider genetic tests for a wider range of conditions to be similarly

problematic. As such, concerns about parent-child relationships may create embedded obligations about genetic testing.

Taken together, existing scholarship shows the complicated ethical and social landscape surrounding prenatal testing for Down syndrome. The notion of clinical utility is shifting from a traditional understanding that medical procedures should have clinically actionable outcomes to one that recognizes the value for personally actionable outcomes, as well. While the dominant framing of disability focuses on medical problems that must be overcome, counter-narratives aim to redirect attention towards discriminatory attitudes and exclusionary practices that impact the lives of people with disabilities negatively. Genetic specialists, public health professionals and bioethicists highlight some of the central values and issues at stake, including autonomy, eugenics, resource allocation, and parental responsibilities. This work explores how this constellation of values, beliefs and assumptions intersect in publicly available health messages about prenatal testing for Down syndrome.

METHODS

At a very basic level, "methods" describe how you do what you do. Because of the interdisciplinary nature of my work, I draw upon an array of disciplines and fields, including epidemiology, philosophy, anthropology, communication, policy analysis, empirical bioethics, community health sciences, and Disability Studies. Some parts of this dissertation rely more heavily on a particular way of doing things to fit appropriately its intended audience. Overall, my dissertation is based upon qualitative research. Many qualitative researchers who explore prenatal testing want to understand attitudes towards testing, why people choose to test or not and their responses, decision-making, or reactions to testing (Browner & Press, 1996; Ginsburg

& Rapp, 1999; Ivry, 2008; Kelly & Farrimond, 2012; Ormond et al., 2003; Pilnick, 2004; Press & Browner, 1997; Rapp, 1999; Taylor, 2000; Vanzwieten, Willems, Knegt, & Leschot, 2006).[6]

My work addresses health communication and education within a broader social context. I am most interested in how this communication shapes expectations about prenatal testing and its clinical utility. Specifically, I am interested in how public representations of disability, abortion, and eugenics influence popular thinking about these expectations and perceived utility. I assume that within the US healthcare system, the emphasis on clinical utility governs most healthcare decisions, especially given this era of evidence-based medicine; however, I acknowledge that to-date there is no empirical evidence to substantiate this claim. Because public understanding of the so-called clinical utility of prenatal testing for Down syndrome reflects underlying values and assumptions that we may not be aware of and ultimately informs our adoption and utilization of such testing, I am interested in how individuals produce and reproduce the language in circulation about potential risks and harms of prenatal testing. At the forefront of my inquiry is the following set of research questions:

- How is prenatal genetic testing for Down syndrome represented in the public sphere?
- How does the language and meanings in use in public texts about the clinical utility of emerging testing methodologies evolve over time?
- How do health messages convey what counts as utility of such tests? How is the meaning of utility expressed across different sources (i.e., policy, newspapers, radio, health education, YouTube videos)?
- How do individuals create meaning about utility that differs from scholarly conversations? How and in what contexts do individuals reproduce values, belief and assumptions evident in existing scholarship?
- If the language-in-use differs significantly across sources or between central groups, then how ought we to craft health education and communication, so that patients might be best and fully informed in their decision-making about such tests?

[6] While by no means an exhaustive list, this scholarship captures some of the breadth of this literature. Worth noting, much of this literature directly focuses on patient-provider communication within the clinical encounter.

As these questions illustrate, my research focuses on the ways in which health communication –broadly understood- shapes our public and private understanding of prenatal genetic testing. In particular, these questions highlight what knowledge or ways of knowing commonly circulate in the public sphere. As such, knowledge creation, formation and exchange are central to my work.

Discourse Tracing

With the intent to provide both grounded and practical insights, this applied research addresses issues of language, power, and context. As a methodological anchor, I utilized discourse tracing, a variant of discourse analysis, which aims to critically explore the sharing of knowledge associated with a social change (LeGreco & Tracy, 2009). Discourse analysis is a qualitative research method that illuminates the ways in which language encapsulates things, like identities, actions, values, feelings and technologies; in this manner, discourse analysis reveals how language-in-use enacts social and cultural perspectives, as well as creates situated meanings (Gee, 2010). At its foundation, discourse analysis involves examining texts with a set of guiding questions, in order to interrogate the use of language in a particular context. Discourse tracing extends beyond this most basic discourse analysis, and it interrogates texts across multiple levels of discourse (LeGreco & Tracy, 2009). I employed discourse tracing to analyze circulating language about prenatal testing; such language evolved as emerging genetic testing methodologies translate from bench research into clinical applications and as different social groups engage and exchange dialogue. My analysis elucidates values and assumptions that create the notion of clinical utility and that interplay in private and public decision-making about testing adoption and utilization. I focused on how the purpose, concerns, harms, risks, and benefits were articulated with special attention towards underlying assumptions about each.

Using discourse tracing as a tool, I illustrate the values that are informing the understanding of clinical utility that fuels the translation of prenatal genetic testing into practice and policy.

As LeGreco & Tracy (2009) articulate, discourse tracing involves four major steps: defining a case, collecting multilevel discourse and managing data, applying structured questions to texts to guide analysis, evaluating the data for practical implications and recommendations.

Defining a Case

LeGreco & Tracy (2009) describe defining a case as identifying a turning point in which significant events reshape the public discourse. Genetic testing has a longstanding history in prenatal care and encompasses many scientific, legal and policies, as well as popular cultural events (See Appendix A). However, not until 1997 did the fundamental scientific knowledge dramatically shift (Lo *et al.*, 1997). His publication, "Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis" (1998), is a substantial turning point, in that it is a culmination of advancements in understanding about fetal DNA circulating in maternal blood and DNA amplification technologies. Private and public conversations about noninvasive prenatal diagnosis heightened since this seminal work. The science itself, however, does not make this publication a turning point. With the undertaking of the Human Genome Project, the public was also arguably more attuned to both genetics research and the ethical, legal, and social implications (ELSI) that such work raised (US Department of Energy Genome Programs, 2011). Furthermore, the passage of the ADA ("Americans with Disabilities Act," 1990) coupled with the development of Disability Studies programs across the United States (Cushing & Smith, 2009) fueled disability rights activism that were now being directed towards scholarly and social issues, like assisted dying, prenatal testing, and growth attenuation. As noninvasive prenatal diagnosis now begins to translate into clinical practice

(American College of Obstetricians and Gynecologists, 2012), we continue to see it as the focal point in the public sphere and in private decision-making.

In many ways, Lo's publication was a culmination of many interesting and intersecting arenas. Because of this, I observe changes in language both before and after this case emerged. This development creates language about cell-free fetal DNA testing. Some refer to this as noninvasive prenatal testing, making a tacit comparison between invasive techniques, like amniocentesis or chorionic villus sampling. Some of the risks and benefits are readily apparent in these word choices (i.e., noninvasive versus invasive), but I capture the subtle differences, as well. The ability to move backward and forward chronologically through the case provides the opportunity to detect nuanced changes in our understanding about the clinical utility of prenatal diagnosis. I also illustrate the contextual circumstances that gave rise to noninvasive prenatal diagnosis and describe how we talk about the clinical utility of such research that rapidly translates into practice.

Collecting and Managing Data

I examined a range of "texts". In this instance, the texts are not all written documents, but more broadly representations of Down syndrome in relationship to prenatal genetic testing. The texts include health policies, radio casts, newspapers, peer-reviewed literature, health education, and YouTube videos. I deliberately sampled texts that reflect biomedical, disability, and mainstream perspectives about prenatal genetic testing. I identified texts with the goal of capturing macro-, meso-, and micro- level discourses over the time period from 1990 to 2013. I organized the texts chronologically, so that I could evaluate them in relationship with key social events that may have triggered changes in the discourse over time.

In order to classify texts, I evaluated the scale of their impact, influence, and/or viewership. Individual opinions and self-published works, such as YouTube videos and public

comments qualified as micro-discourse. Health education and peer-reviewed biosciences literature related to Down syndrome incidence qualified as meso-discourse. Texts with a broad and/or unspecified public audience, such as policy or newspapers, qualified as macro-discourse.

Applying Structured Questions

As LeGreco and Tracy (2009) describe, structured questions are different from research questions. They are informed by the background literatures, in order to identify themes in a qualitative data set. The specific questions I have used for my analysis are listed in Table 2. This set of questions provided a detailed framework to understand and to analyze the texts.

Evaluating Data

The final step involves evaluating the data, taking into account both theoretical and practical applications.[7] While prenatal testing for Down syndrome is the case explicitly studied in this analysis, as part of the theoretical evaluation, I discuss how the insights about language and power from this case analysis might extend to conversations about prenatal testing for other genetic conditions, to conversations about up-to-date, accurate, high-quality health education, and to broader conversations about reproductive rights. As part of the practical evaluation, I provide a general set of recommendations. These recommendations address areas for future research and otherwise how to improve current scholarly conversations. However, more importantly, I provide recommendations for the key stakeholders who I have identified throughout the research process. Namely, I discuss mechanisms for how people with disabilities might talk about and frame their arguments about prenatal testing for specific genetic conditions to health professionals and the general public; I highlight key issues and strategies for solutions to those involved with creating health education materials on the topic; I also note specific

[7] Chapter 6 overviews the specific recommendations, looking across all data sources.

language and topics that providers involved with the offer of such testing should consider in clinical practice, in order to improve patient-provider communication.

Table 2. Analysis guide

Guiding Questions
Stakeholders
• What key actors are involved in the discourse? • What disciplinary discourses enter the conversation? • Who is represented in the discourse?
Situated meaning
• How do word-specific choices create particular meanings?
<u>Cell-free fetal DNA testing</u>
• How is testing discussed? • What benefits/concerns are expressed? Who expresses them? • How is this testing distinguished from invasive diagnostic tests? • How is this testing distinguished from noninvasive screening tests?
<u>Clinical utility</u>
• How is clinical utility of noninvasive prenatal testing expressed? • What is the purpose(s) of such testing? • What counts as clinical utility, when not explicitly discussed?
<u>Down syndrome</u>
• What models of disability dominate the discourse? • How is the medical or social model of disability expressed? • Do other models of disability enter the conversation?
<u>Eugenics</u>
• How is eugenics discussed? • How do different actors talk about it? • What actors silence eugenics talk?

Guiding Questions

Choice

- How is choice used?
- What choices are being articulated or discussed?
- How do different actors talk about it?
- What actors silence choice talk?

References

Alderson, P. (2001). Down's syndrome: cost, quality and value of life. *Social Science & Medicine (1982), 53*(5), 627-638.

American College of Obstetricians and Gynecologists. (2007a). Screening for Fetal Chromosomal Abnormalities. *Obstet Gynecol, 109*(1).

American College of Obstetricians and Gynecologists. (2007b). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

American College of Obstetricians and Gynecologists. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm

American College of Obstetricians and Gynecologists. (2007d). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. *Obstet Gynecol, 110*(6), 1459.

American College of Obstetricians and Gynecologists. (2012). Noninvasive prenatal testing for fetal aneuploidy. *Obstet Gynecol, 120*(545), 1532–1534.

Americans with Disabilities Act, Pub. L. No. 110-325 (1990).

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.

Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Asch, A. (2003). Disability equality and prenatal testing: contradictory or compatible? *Florida State University Law Review. Florida State University. College Of Law, 30*(2), 315-342.

Baily, M. A., & Murray, T. H. (2008). Ethics, evidence, and cost in newborn screening. *Hastings Center Report, 38*(3), 23-31.

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? *The American Journal of Bioethics, 9*(8), 48-56.

Biesecker, B. B., & Hamby, L. (2000). What difference the disability community arguments should make for the delivery of prenatal genetic information. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Bindra, R., Heath, V., Liao, A., Spencer, K., & Nicolaides, K. H. (2002). One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. *Ultrasound in Obstetrics and Gynecology, 20*(3), 219-225. doi: 10.1046/j.1469-0705.2002.00808.x

Botkin, J. R., Clayton, E. W., Fost, N. C., Burke, W., Murray, T. H., Baily, M. A., . . . Ross, L. F. (2006). Newborn Screening Technology: Proceed With Caution. *Pediatrics, 117*(5), 1793-1799. doi: 10.1542/peds.2005-2547

Browner, C. H. (2000). Situating Women's Reproductive Activities. *American Anthropologist, 102*(4), 773-788.

Browner, C. H., & Press, N. (1996). The production of authoritative knowledge in American prenatal care. *Medical Anthropology Quarterly, 10*(2), 141-156.

Browner, C. H., & Press, N. A. (1995). The normalization of prenatal diagnostic screening. *Conceiving the new world order: The global politics of reproduction*, 307-322.

Buchanan, A., Brock, D. W., Daniels, N., & Wikler, D. (2000). *From Chance to Choice: Genetics and Justice*. Cambridge: Cambridge Univ Pr.

Burke, W., Atkins, D., Gwinn, M., Guttmacher, A., Haddow, J., Lau, J., . . . Wiesner, G. L. (2002). Genetic Test Evaluation: Information Needs of Clinicians, Policy Makers, and the Public. *American Journal of Epidemiology, 156*(4), 311-318. doi: 10.1093/aje/kwf055

Chao, S., Roberts, J. S., Marteau, T. M., Silliman, R., Cupples, L. A., & Green, R. C. (2008). Health behavior changes after genetic risk assessment for Alzheimer disease: The REVEAL Study. *Alzheimer disease and associated disorders, 22*(1).

Childress, J. F., Faden, R. R., Gaare, R. D., Gostin, L. O., Kahn, J., Bonnie, R. J., . . . Nieburg, P. (2002). Public health ethics: mapping the terrain. *The Journal of Law, Medicine & Ethics, 30*(2), 170-178.

Constantin, C. M., Faucett, A., & Lubin, I. M. (2005). A primer on genetic testing. *Journal of Midwifery & Women's Health, 50*(3), 197-204.

Cushing, P., & Smith, T. (2009). Multinational Review of English Language Disability Studies Degrees and Courses. *Disability Studies Quarterly, 29*(3).

Davis, D. S. (2010). *Genetic dilemmas : reproductive technology, parental choices, and children's futures*. Oxford; New York: Oxford University Press.

Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. *American Journal of Obstetrics and Gynecology, 191*(3), 1044-1048.

Gee, J. P. (2010). *An introduction to discourse analysis : theory and method*. New York [etc.]: Routledge.

Ginsburg, F., & Rapp, R. (1999). Fetal reflections: Confessions of two feminist anthropologists as mutual informats. In L. M. Morgan & M. W. Michaels (Eds.), *Fetal subjects, feminist positions*. Philadelphia: University of Pennsylvania Press.

Green, R. C., Roberts, J. S., Cupples, L. A., Relkin, N. R., Whitehouse, P. J., Brown, T., . . . Group, R. S. (2009). Disclosure of APOE genotype for risk of Alzheimer's disease. *The New England journal of medicine, 361*(3), 245-254.

Groce, N. E. (1985). *Everyone here spoke sign language : hereditary deafness on Martha's Vineyard*. Cambridge, Mass.: Harvard University Press.

Haller, B. A. (2010). *Representing disability in an ableist world : essays on mass media*. Louisville, KY: The Advocado Press.

Ivry, T. (2008). At the Back Stage of Prenatal Care: Japanese Ob-Gyns Negotiating Prenatal Diagnosis. *Medical Anthropology Quarterly, 20*(4), 441-468.

Kalfoglou, A., Suthers, K., Scott, J., & Hudson, K. (2004). Reproductive genetic testing: What America thinks. *Washington, DC: Genetics and Public Policy Center*.

Kelly, S., & Farrimond, H. (2012). Non-invasive prenatal genetic testing: a study of public attitudes. *Public Health Genomics, 15*(2), 73-81.

Khoury, M., McBride, C., Schully, S., Ioannidis, J. P., Feero, W. G., Janssens, A. C. J., . . . Cargill, M. (2009). The scientific foundation for personal genomics: recommendations from a National Institutes of Health–Centers for Disease Control and Prevention multidisciplinary workshop. *Genetics in Medicine, 11*(8), 559.

Khoury, M. J., Jones, K., & Grosse, S. D. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. *Genetics in Medicine, 8*(3), 191.

Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: conversations with my son. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. *Science translational medicine, 4*(137), 137ra176-137ra176.

Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". *AJOB Primary Research, 2*(2), 8-20.

Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing. *Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70*(1), 7-9.

LaRusse, S., Roberts, J. S., Marteau, T. M., Katzen, H., Linnenbringer, E. L., Barber, M., . . . Relkin, N. R. (2005). Genetic susceptibility testing versus family history-based risk assessment: Impact on perceived risk of Alzheimer disease. *Genetics in Medicine, 7*(1), 48-53.

LeGreco, M., & Tracy, S. J. (2009). Discourse tracing as qualitative practice. *Qualitative Inquiry, 15*(9), 1516-1543.

Leung, T. Y., Chan, L. W., Law, L. W., Sahota, D. S., Fung, T. Y., Leung, T. N., & Lau, T. K. (2009). First trimester combined screening for Trisomy 21 in Hong Kong: outcome of the first 10,000 cases. *The Journal of Maternal-Fetal & Neonatal Medicine, 22*(4), 300-304. doi: 10.1080/14767050802430842

Lippman, A. (1999). Embodied knowledge and making sense of prenatal diagnosis. *Journal of genetic counseling, 8*(5), 255-274.

Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. *American Journal of Human Genetics, 51*(4), 936.

Lo, Y., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W., & Wainscoat, J. S. (1997). Presence of fetal DNA in maternal plasma and serum. *The Lancet, 350*(9076), 485-487.

Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. *The American Journal of Human Genetics, 62*(4), 768-775.

Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: a commentary. *American journal of medical genetics. Part A, 155A*(8), 1777-1785.

Malone, F. D., Canick, J. A., Ball, R. H., Nyberg, D. A., Comstock, C. H., Bukowski, R., . . . D'Alton, M. E. (2005). First-Trimester or Second-Trimester Screening, or Both, for Down's Syndrome. *New England Journal of Medicine, 353*(19), 2001-2011. doi: doi:10.1056/NEJMoa043693

Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. *Prenatal diagnosis, 19*(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Markel, H. (1997). Scientific advances and social risks: historical perspectives of genetic screening programs for Sickle Cell Disease, Tay-Sachs Disease, neural tube defects and Down Syndrome, 1970-1997. *Medical and Public Health Law*. http://biotech.law.lsu.edu/research/fed/tfgt/appendix6.htm

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. *Sociology of health & illness, 32*(1), 37-56.
Markens, S., Browner, C. H., & Press, N. A. (1999). Because of the risks': how US pregnant women account for refusing prenatal screening. *Social Science & Medicine, 49*(3), 359-369.
Marteau, T., Drake, H., & Bobrow, M. (1994). Counselling following diagnosis of a fetal abnormality: the differing approaches of obstetricians, clinical geneticists, and genetic nurses. *Journal of medical genetics, 31*(11), 864-867.
Marteau, T., Plenicar, M., & Kidd, J. (1993). Obstetricians presenting amniocentesis to pregnant women: practice observed. *Journal of Reproductive and Infant Psychology, 11*(1), 3-10.
Miron-Shatz, T., Hanoch, Y., Graef, D., & Sagi, M. (2009). Presentation Format Affects Comprehension and Risk Assessment: The Case of Prenatal Screening. *Journal of Health Communication, 14*(5), 439-450. doi: 10.1080/10810730903032986
Murkoff, H. E., & Mazel, S. (2008). *What to expect when you're expecting* (4th ed.). New York: Workman Pub.
Nadler, H. L. (1968). Antenatal detection of hereditary disorders. *Pediatrics, 42*(6), 912-918.
National Coalition for Health Professional Education in Genetics, & National Society of Genetic Counselors. (2012). Prenatal Screening/Testing Options for Trisomy 21 and Other Fetal Aneuploidies. In N.-I. P. T. N. Factsheet (Ed.).
Nelson, J. L. (1998). The meaning of the act: reflections on the expressive force of reproductive decision making and policies. *Kennedy Inst Ethics J, 8*(2), 165-182.
Nicolaides, K. H. (2011). Screening for fetal aneuploidies at 11 to 13 weeks. *Prenatal diagnosis, 31*(1), 7-15. doi: 10.1002/pd.2637
Nicolaides, K. H., Spencer, K., Avgidou, K., Faiola, S., & Falcon, O. (2005). Multicenter study of first-trimester screening for trisomy 21 in 75 821 pregnancies: results and estimation of the potential impact of individual risk-orientated two-stage first-trimester screening. *Ultrasound in Obstetrics and Gynecology, 25*(3), 221-226. doi: 10.1002/uog.1860
Norton, M. E., Brar, H., Weiss, J., Karimi, A., Laurent, L. C., Caughey, A. B., . . . Song, K. (2012). Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18. *American Journal of Obstetrics and Gynecology, 207*(2), 137.e131-137.e138. doi: http://dx.doi.org/10.1016/j.ajog.2012.05.021
Oliver, M. (1990). *The individual and social models of disability*. Paper presented at the Joint Workshop of the Living Options Group and the Research Unit of the Royal College of Physicians.
Oliver, M. (1996). *Defining Impairment and Disability: Issues at Stake.* Paper presented at the Exploring the Divide, University of Leeds.
Ormond, K. E., Gill, C. J., Semik, P., & Kirschner, K. L. (2003). Attitudes of health care trainees about genetics and disability: Issues of access, health care communication, and decision making. *Journal of genetic counseling, 12*(4), 333-349.
Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine, 13*(11), 913-920.

Pilnick, A. (2004). 'It's Just One of the Best Tests that We've Got at the Moment': the Presentation of Nuchal Translucency Screening for Fetal Abnormality in Pregnancy. *Discourse & Society, 15*(4), 451-465. doi: 10.1177/0957926504043710
Press, N., & Browner, C. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.
Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.
Press, N., Wilfond, B., Murray, M., & Burke, W. (2011). The power of knowledge: How carrier and prenatal screening altered the clinical goals of genetic testing. In W. Burke, K. A. Edwards, S. Goering, S. Holland & S. B. Trinidad (Eds.), *Achieving Justice in Genomic Translation: Re-Thinking the Pathway to Benefit: Re-Thinking the Pathway to Benefit*: Oxford University Press, USA.
Rapp, R. (1993). Amniocentesis in sociocultural perspective. *Journal of genetic counseling, 2*(3), 183-196.
Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), *Representations of motherhood*: Yale University Press.
Rapp, R. (1998). Refusing prenatal diagnosis: The meanings of bioscience in a multicultural world. *Science, technology & human values, 23*(1), 45-70.
Rapp, R. (1999). *Testing women, testing the fetus: The social impact of amniocentesis in America*.
Riley, C. A. (2005). *Disability and the media: prescriptions for change*. Hanover, NH: University Press of New England.
Roberts, J. S., Barber, M., Brown, T. M., Cupples, L. A., Farrer, L. A., LaRusse, S. A., . . . Green, R. C. (2004). Who seeks genetic susceptibility testing for Alzheimer's disease? Findings from a multisite, randomized clinical trial. *Genetics in Medicine, 6*(4).
Rothman, B. K. (1994). The Tentative Pregnancy: Then and Now. In K. H. Rothenberg & E. J. Thomson (Eds.), *Women and prenatal testing : facing the challenges of genetic technology*. Columbus: Ohio State University Press.
Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), *The disability studies reader* (2nd ed., pp. xviii, 451 p.). New York: Routledge.
Secretary's Advisory Committee on Genetic Testing. (2000). Enhancing the oversight of genetic tests: recommendations of the SACGT. from http://oba.od.nih.gov/oba/sacgt/highlights/sacgt012700summ.pdf
Shapiro, J. P. (1993). *No pity : people with disabilities forging a new civil rights movement*. New York: Times Books.
Skotko, B. G. (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? *Archives of disease in childhood, 94*(11), 823-826.
Spencer, K., Souter, V., Tul, N., Snijders, R., & Nicolaides, K. H. (1999). A screening program for trisomy 21 at 10–14 weeks using fetal nuchal translucency, maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A. *Ultrasound in Obstetrics and Gynecology, 13*(4), 231-237. doi: 10.1046/j.1469-0705.1999.13040231.x
Steinbock, B. (2000). Disability, Prenatal Testing, and Selective Abortion. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Stern, A. M. (2005). Sterilized in the name of public health: race, immigration, and reproductive control in modern California. *American Journal of Public Health, 95*(7), 1128.

Task Force on Genetic Testing, Holtzman, N. A., & Watson, M. S. (1998). *Promoting safe and effective genetic testing in the United States : final report of the Task Force on Genetic Testing*. Baltimore: Johns Hopkins University Press.

Taylor, J. S. (2000). Of sonograms and baby prams: Prenatal diagnosis, pregnancy, and consumption. *Feminist Studies, 26*(2), 391-418.

Ten Great Public Health Achievements--United States, 1900-1999. (1999). *JAMA: The Journal of the American Medical Association, 281*(16), 1481-1481. doi: 10.1001/jama.281.16.1481

US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml

Vanzwieten, M., Willems, D., Knegt, L., & Leschot, N. (2006). Communication with patients during the prenatal testing procedure: An explorative qualitative study. *Patient Educ Couns, 63*(1-2), 161-168. doi: 10.1016/j.pec.2005.09.013

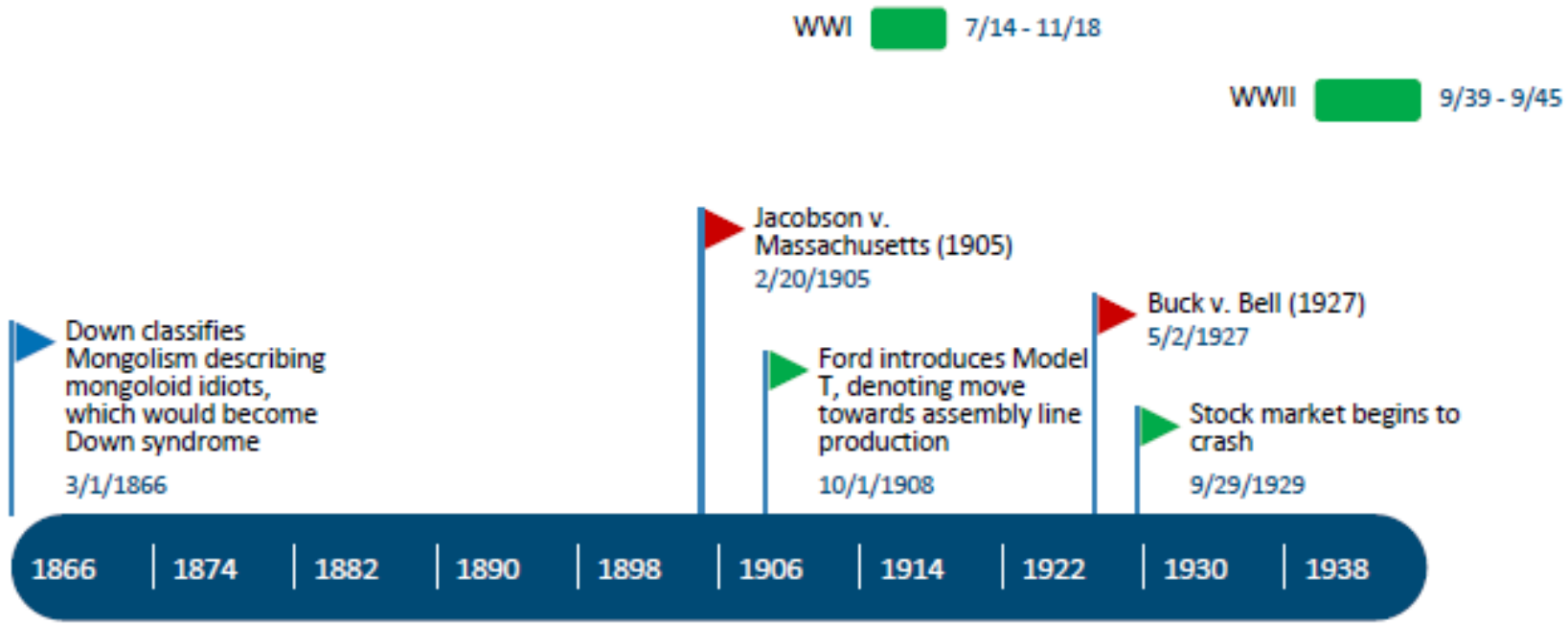
Wald, N. J., Huttly, W. J., & Hackshaw, A. K. (2003). Antenatal screening for Down's syndrome with the quadruple test. *The Lancet, 361*(9360), 835-836. doi: http://dx.doi.org/10.1016/S0140-6736(03)12680-3

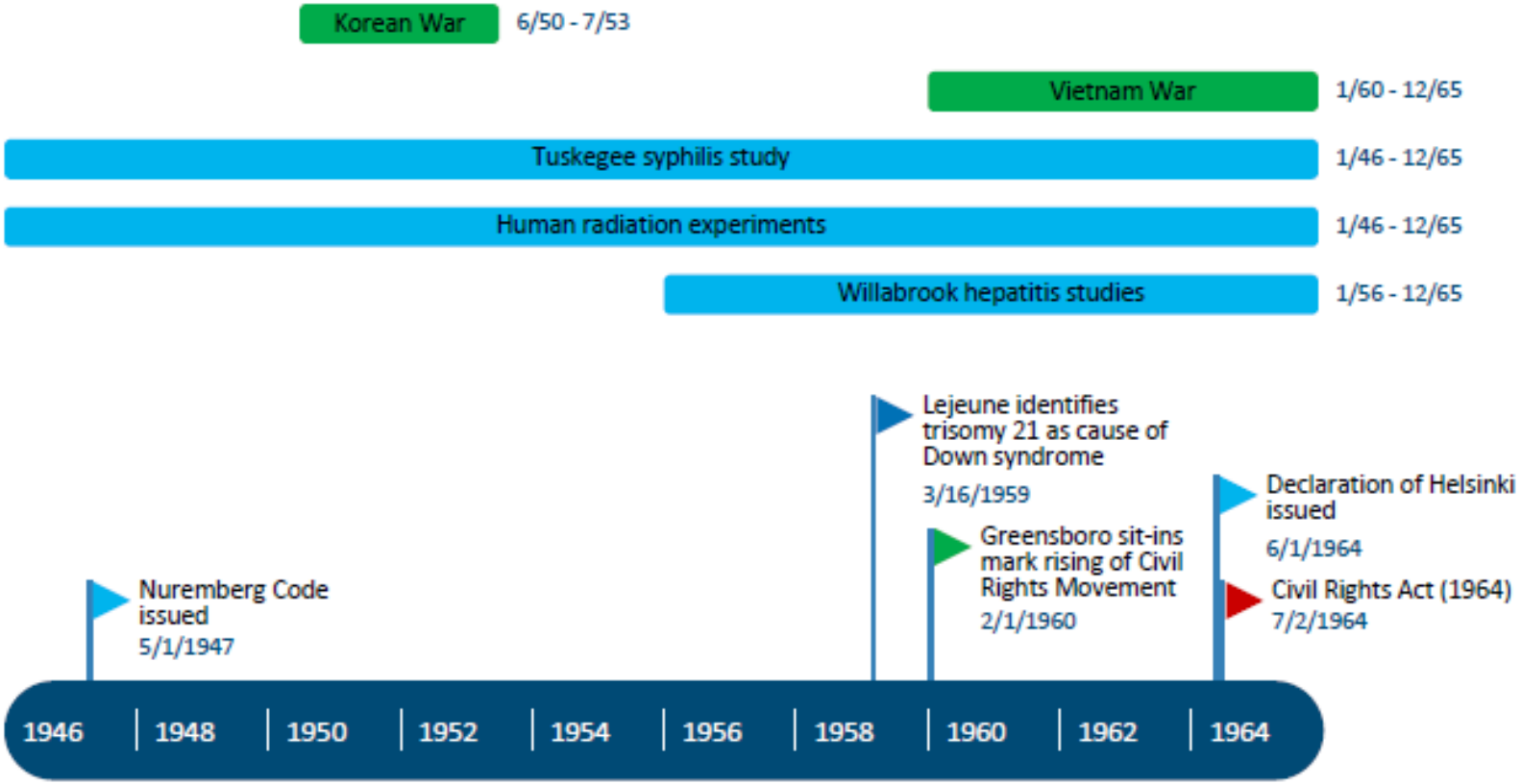
Wald, N. J., Huttly, W. J., Murphy, K. W., K, A., Bestwick, J. P., & Rodeck, C. H. (2009). Antenatal screening for Down's syndrome using the Integrated test at two London hospitals. *Journal of Medical Screening, 16*(1), 7-10. doi: 10.1258/jms.2009.008094

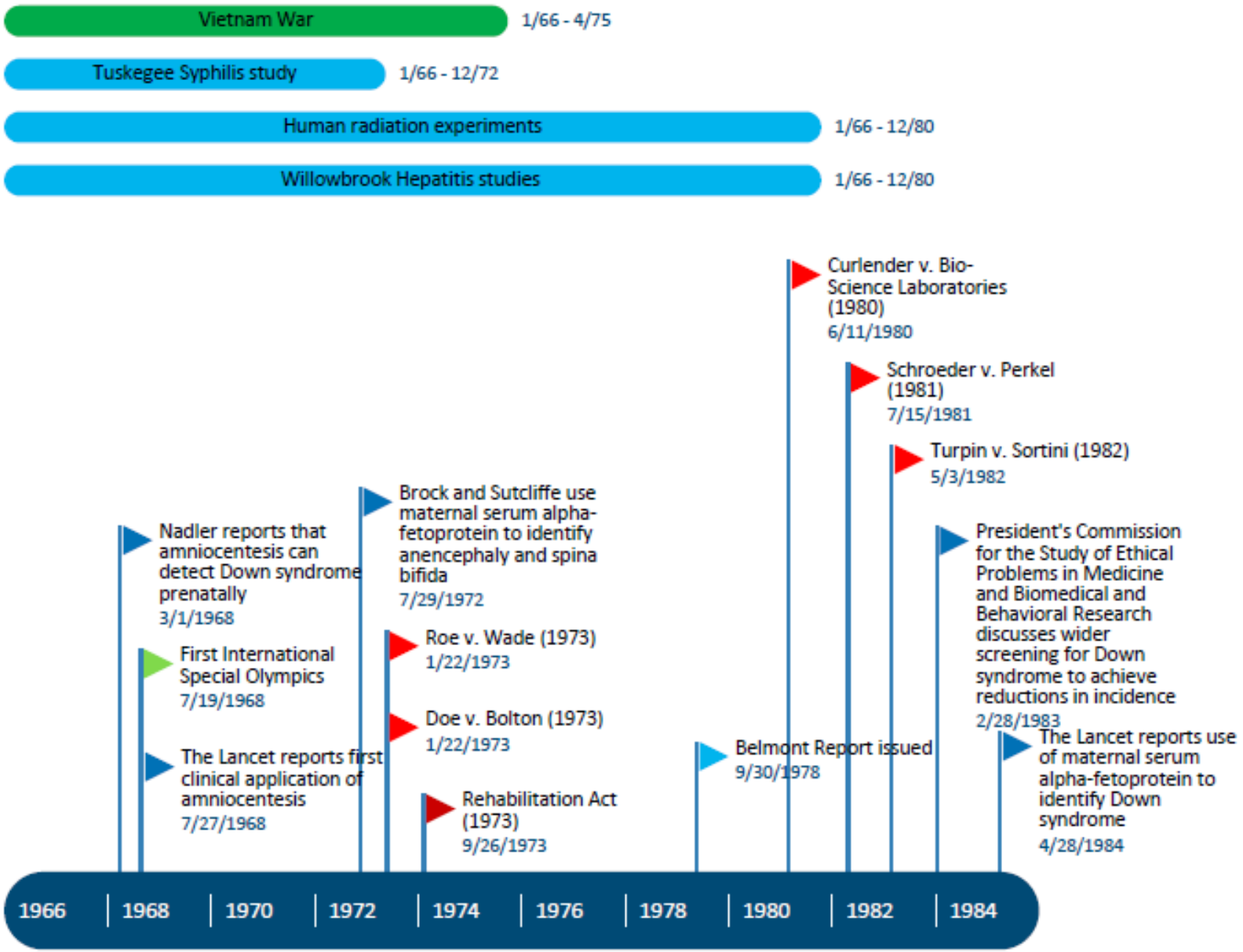
Zick, C. D., Mathews, C. J., Roberts, J. S., Cook-Deegan, R., Pokorski, R. J., & Green, R. C. (2005). Genetic Testing For Alzheimer's Disease And Its Impact On Insurance Purchasing Behavior. *Health Affairs, 24*(2), 483-490. doi: 10.1377/hlthaff.24.2.483

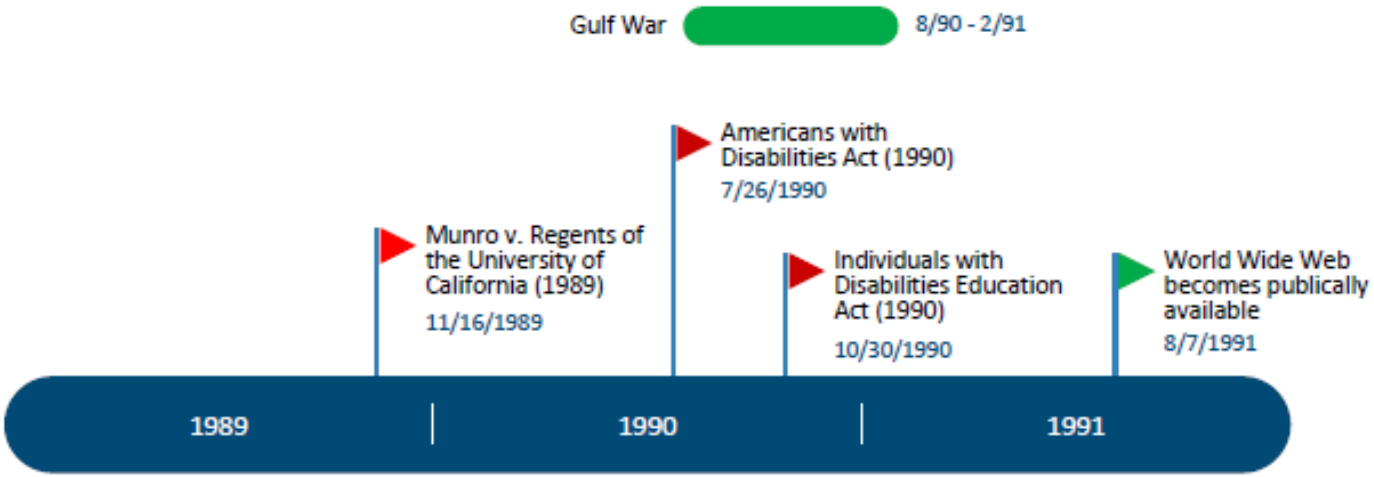
Zimmern, R. L., & Kroese, M. (2007). The evaluation of genetic tests. *Journal of Public Health, 29*(3), 246-250. doi: 10.1093/pubmed/fdm028

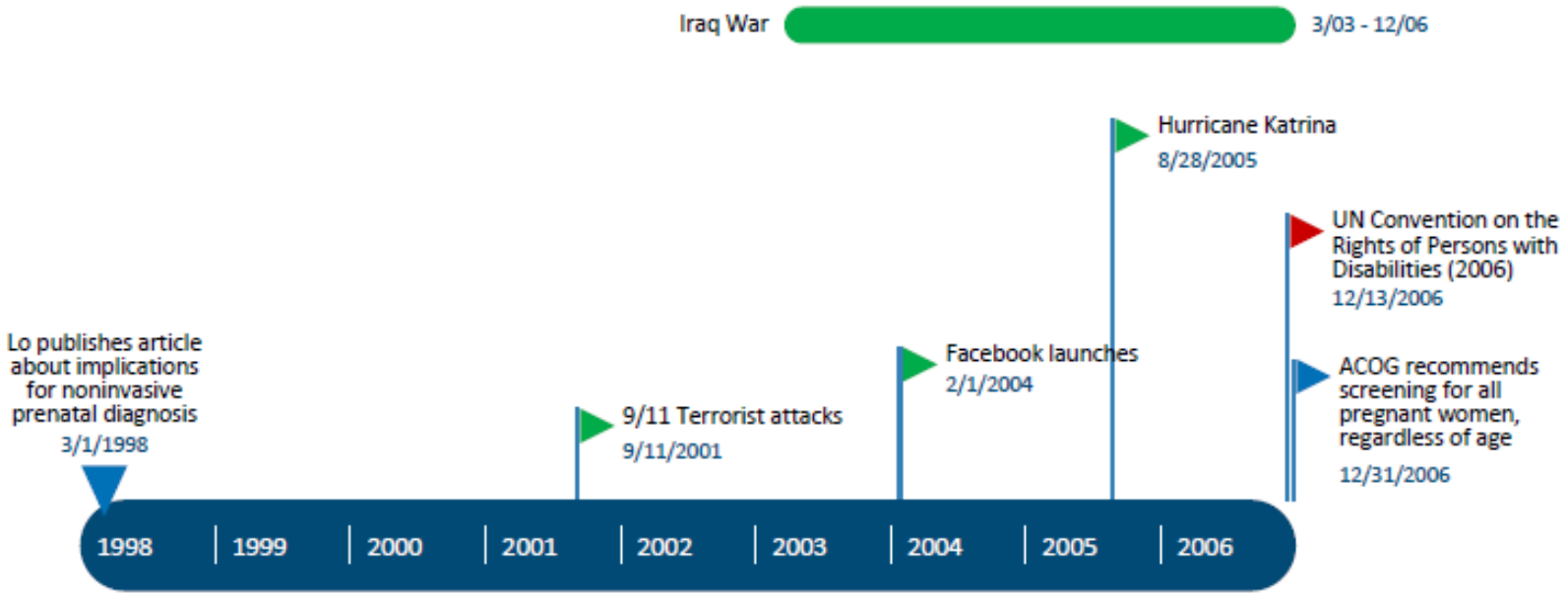
Appendix A. Timeline of key socio-cultural events




Korean War
6/50 - 7/53
Vietnam War
1/60 - 12/65
Tuskegee syphilis study
1/46 - 12/65
Human radiation experiments
1/46 - 12/65
Willabrook hepatitis studies
1/56 - 12/65
Lejeune identifies trisomy 21 as cause of Down syndrome
3/16/1959
Declaration of Helsinki issued
6/1/1964
Greensboro sit-ins mark rising of Civil Rights Movement
2/1/1960
Civil Rights Act (1964)
7/2/1964
Nuremberg Code issued
5/1/1947
1946
1948
1950
1952
1954
1956
1958
1960
1962
1964


Vietnam War
1/66 - 4/75
Tuskegee Syphilis study
1/66 - 12/72
Human radiation experiments
1/66 - 12/80
Willowbrook Hepatitis studies
1/66 - 12/80
Curlender v. Bio-Science Laboratories (1980)
6/11/1980
Schroeder v. Perkel (1981)
7/15/1981
Turpin v. Sortini (1982)
5/3/1982
Brock and Sutcliffe use maternal serum alpha-fetoprotein to identify anencephaly and spina bifida
7/29/1972
Nadler reports that amniocentesis can detect Down syndrome prenatally
3/1/1968
Roe v. Wade (1973)
1/22/1973
First International Special Olympics
7/19/1968
Doe v. Bolton (1973)
1/22/1973
The Lancet reports first clinical application of amniocentesis
7/27/1968
Rehabilitation Act (1973)
9/26/1973
Belmont Report issued
9/30/1978
President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research discusses wider screening for Down syndrome to achieve reductions in incidence
2/28/1983
The Lancet reports use of maternal serum alpha-fetoprotein to identify Down syndrome
4/28/1984
1966
1968
1970
1972
1974
1976
1978
1980
1982
1984


Gulf War
8/90 - 2/91
Americans with Disabilities Act (1990)
7/26/1990
Munro v. Regents of the University of California (1989)
11/16/1989
Individuals with Disabilities Education Act (1990)
10/30/1990
World Wide Web becomes publically available
8/7/1991
1989
1990
1991


Iraq War
3/03 - 12/06
Hurricane Katrina
8/28/2005
UN Convention on the Rights of Persons with Disabilities (2006)
12/13/2006
Lo publishes article about implications for noninvasive prenatal diagnosis
3/1/1998
Facebook launches
2/1/2004
9/11 Terrorist attacks
9/11/2001
ACOG recommends screening for all pregnant women, regardless of age
12/31/2006
1998
1999
2000
2001
2002
2003
2004
2005
2006

Policy provides the backdrop for our conversations about prenatal genetic testing. In this arena, several intersecting policies help to build the narrative: national policies, including abortion policies and healthcare coverage policies; state policies, which implement the aforementioned national policies, as well as create legal precedent for wrongful birth and wrongful life tort; professional policies, which govern the healthcare providers involved with the offer, testing, and follow-up care; advocacy organization policies that aim to shape healthcare practices and inform public policies.

In this section, I illuminate the rhetorical devices that the disability and medical communities use in policy statements to frame the issues surrounding prenatal genetic screening and testing. This qualitative analysis focuses on how these stakeholder groups imitate and engage the national debate about abortion policy in both form and content, particularly in their use of language about choice.

Chapter 2 POLICY

In the public sphere, the abortion debate has co-evolved with debates over prenatal testing, as genetic testing for Down syndrome entered clinical practice about the same time as North America began questioning the legality of abortion (Nadler, 1968; "Roe v. Wade," 1973). Historically, amniocentesis was offered to women over the age of 35, chosen because the perceived benefit of knowing the fetal karyotype outweighs the risk of pregnancy loss and the risk of having a child with Down syndrome increases with age. Nonetheless, the procedure has some risk of pregnancy loss, or limb and digit loss for the fetus (Eddleman *et al.*, 2006). Since the 1970s, technological advances have reduced these associated risks, and new medical procedures allow for similar testing using a maternal blood sample (Lo *et al.*, 1998). For some, the results of such testing may shape decisions about pregnancy management – that is, whether to continue or terminate a pregnancy following prenatal diagnosis.

Arguments in favor of and in opposition to the availability of prenatal genetic testing have focused on women's reproductive choice and ultimately fallen into the discursive strategies used in the ongoing public conversation about abortion. Many pro-life groups object to abortion on moral and philosophical grounds, understanding the fetus as a person, and therefore affording

rights to a fetus that the State should protect; conversely, pro-choice advocates argue that a pregnant woman's rights trump any that might be afforded to a fetus. This polarized debate raises questions about whether abortion should be legally allowed and whether public funds ought to support such services.

The moral and political issues related to abortion can become distractions from the alternative, yet pressing question of whether prenatal genetic screening and testing should be offered, and if so, how and what procedures should be offered. They also raise related questions about who ought to be offered such testing, when, for what purpose, and for whose benefit. At the intersections of clinical practice and public health, such questions bring to the foreground broader issues about access to healthcare services, as well as social expectations about family and parenting.

As Varvasovszky & Brugha (2000) describe in relationship to health policy and planning, stakeholders are actors who have an interest, are affected, or can influence a particular issue. Previous scholarship has looked at the bioethical and rhetorical dimensions of prenatal genetic testing (Lippman & Wilfond, 1992; Potter *et al.*, 2008; Press & Browner, 1994; Seavilleklein, 2009; Wilkinson, 2008), but the focus has been on prospective parents as the key stakeholder. This paper extends this conversation by taking into consideration the disability and medical communities as a key stakeholders of interest. Yet, a range of individuals make up both of these communities. For example, the disability community includes: people with disabilities (both with and without prenatally identifiable conditions), their families, disability rights activists and their allies. On the other side, the medical community includes: physicians (both those with specialized genetic training and those without), allied health professionals, healthcare policymakers, healthcare insurers and payers.

I select these stakeholders in efforts to highlight their arguably unique vantage points and power positions. While both share interest in prenatal testing and related policy will impact both, the disability community has less power in decision-making than the medical community. Unlike the medical community, the disability community only has access to prospective parents if the medical community refers patients to people with disabilities and their families. Clinical practice guidelines inform practice and policies, including how and which services health insurance covers. In contrast, the disability communities' position statements have relatively limited impact: insofar as clinicians read them and integrate them into policies and/or practice, or as prospective parents read them and are compelled to take personal action. The disability and medical communities hold seemingly oppositional positions: the medical community supports the practice, while the disability community opposes it. However, given the diversity within each group, these communities' policy statements must reflect a variety of positions related to prenatal testing. That is to say, while it may broadly be true that members of disability communities are concerned about the practice of prenatal testing, not all would seek to eliminate the practice altogether, and like the general community, some may object due to other moral concerns. In contrast, medical communities have specific professional duties and obligations to their patients, and I would expect these communities to have similar variety in terms of their perspectives about issues that prenatal testing, especially given that pro-life ideology and critiques about the practice of prenatal testing are present within these communities. This embedded diversity of attitudes and opinions, particularly as projected to a public audience, is central to my analysis – as they highlight nuance and "greyness" in an often polarized debate about abortion.

This section explores how these key stakeholders use the language of choice. This analysis shows that both the disability and medical communities trade on this language to subtly persuade their readers, particularly those who may hold differing political and ethical positions. By evoking multiple meanings of choice in their statements, they promote their ideological perspectives in the public sphere. The disability communities frame the issues at stake through a lens of "informed choice", and the medical communities use a healthcare consumerism approach, suggesting that patients have a range of options to choose as they wish. These deliberate word selections build upon the ever-present, yet tacit conversation about abortion. The analysis identifies rhetorical strategies that these stakeholders should implement in policy statements, given that both emerging technologies and current practice may warrant new guidance.

METHODS

I examine the debate between the disability and medical communities through a purposive sample of policy statements. Both the disability and medical communities represent stakeholders in the evolving policy related to prenatal genetic screening and testing. Disability communities are diverse with regards to political and religious viewpoints and are not explicitly asking whether or not abortion should be legal, but rather whether individuals ought to be able to choose the traits of a particular fetus, or more precisely – given current technologies – to choose against certain traits. As a contrast, I have selected the medical community, because it remains the gatekeeper of both prenatal testing and pregnancy termination services and is a valued profession with political power.

By design, policy statements represent a collective of opinions for their respective groups. I make no assertion that such texts provide a representative account of all people with disabilities or all medical professionals. Nonetheless, these documents convey a specific point-

of-view. These texts are not direct responses to each other; in fact, the disability community's statement likely does not intend to counter that of the medical community or vice versa (e.g., the disability communities may have never read clinical practice guidelines about prenatal testing, or alternatively, the authors of the clinical practice guidelines may have never heard the messages of the disability communities). Given that these statements are displayed publicly and available to a general public audience, I suspect that the messages they convey are insulated from each other, rather than truly shared and exchanged. For the most part, the public gains access to these policy statements indirectly, filtered through the media or in how they inform clinical practices that impact patient care.

Disabled Persons International

To explore the demands of disability communities in relation to genetic testing, I use the Disabled Persons International (DPI) policy statement entitled "Disabled People Speak on the New Genetics" (2000). DPI is a network of disabled persons' organizations that aims to promote the human rights of disabled persons by encouraging economic and social integration, representing constituents from 130 countries (Disabled People's International, 2012). I selected this organization because of its cross-disability support, as opposed to a disability-specific organization, such as Global Down Syndrome Foundation. While the topic of "new genetics" addresses issues related to genetic research and genetic technologies generally, the text provides specific language regarding prenatal screening and testing. Because this text provides little explication as to why the provided demands are justifiable, I supplement this primary text with the work from disability rights advocates and Disability Studies scholarship. As a Feminist Disability Studies scholar, Asch (1999) critiques the ways in which prenatal testing is offered in current practice, arguing that in order to protect and promote reproductive autonomy, structures

must ensure not only access to legal abortion, but also informed consent about reproductive options.. In other words, the Disability Studies critique of prenatal testing is distinct from a pro-life stance; in fact, many Disability Studies scholars advocate for reproductive autonomy, including the access to abortion services for all women.

American Congress of Obstetricians and Gynecologists

In order to see how the medical community has addressed the demands of disability communities, I use the American Congress of Obstetricians and Gynecologists (ACOG) news releases related to this topic entitled "ACOG's Screening Guidelines on Chromosomal Abnormalities: What They Mean to Patients and Physicians" (ACOG Office of Communications, 2007a) and "New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women" (ACOG Office of Communications, 2007c). ACOG is a nonprofit advocacy organization comprised of women's healthcare physicians. I chose to examine this web-based communication as a primary text because of its accessibility to the public, supplementing these texts with the practice guideline only available to members and those with subscriptions to the organization's journal, Obstetrics & Gynecology.

It is worth noting that medical professionals with specialized training in genetics contributed to developing both sets of clinical practice guidelines. As Davis (2010) notes, genetic specialists – perhaps even more than other health professionals, including obstetricians and gynecologists – hold autonomy in high regard. Davis suggests that this focus is at least in part due to the fact that the medical genetics community understands its historical connection to eugenics. Within the field, the focus on patient autonomy has led to the development of two central tenets: value-neutrality and nondirective counseling. Taken together, the value of a decision is solely based upon what is right for that particular patient and providers should not try

to influence a decision (Davis, 2010). We might expect as part of their own professionalism that obstetricians and gynecologists value patient autonomy and perhaps patient-centered care; however, given that their training likely focuses on promoting healthy pregnancies and birth defect prevention, they may be less influenced or committed to value-neutrality and nondirective counseling that those with specialized genetics training. Interestingly, the authors of these guidelines all have advanced training in genetics, highlighting a potential professional tension and/or trade-off that may be made to balance health promotion activities with nondirective counseling.

With the intent to provide both grounded and practical insights, this research addresses issues of language, power, and context. Discourse analysis is a qualitative research method that illuminates the ways in which language encapsulates things, like identities, actions, and values, revealing how language enacts social perspectives and creates situated meanings (Gee, 2010). By examining texts with a set of guiding questions to interrogate the use of language, the analysis employed a form of directed content analysis (Hsieh & Shannon, 2005). Using ATLAS.ti 7™, the analysis explored how the disability and medical communities' statements differentiate their interests and claims from the abortion debate.

Findings and Interpretation

Extending Choice

Within North America between 1960 and 1985, the public discourse –in media, in popular vocabulary, medical practice, and law− articulated both motherhood and abortion as a "choice" (Condit, 1990). The language of choice surfaces in these texts about prenatal genetic screening and testing; however, the stakeholders extend the meaning of choice, embedding choice within larger conversations about autonomy and consumerism.

DPI appeals to an expanded conceptualization of choice. DPI describes prenatal testing as lacking "informed choice" and suggesting "no free choice" can occur within the current medical context (p. 6). This understanding of choice is implicitly connected to bioethical understandings about autonomy, which outline that voluntariness, understanding, recommendation, and authorization are some of the key elements of informed consent (Beauchamp & Childress, 1994). Consistent with other Disability Studies scholarship (for example, Asch & Wasserman, 2005; Bérubé, 1998; Saxton, 2006), DPI suggests that medical practice fails to provide complete and accurate information about disability (understanding) and that society offers inadequate social supports for many families to raise a child with a disability (voluntariness). While bioethics specifically uses the term informed consent, DPI substitutes choice in place of consent:

> With respect to the impact of genetics on reproduction, we support women's right to choose with respect to their pregnancies. However, we deplore the context in which these choices are made. There is no informed choice as long as genetic counseling is directive and misinforms parents about the experience of disability. There can be no free choice as long as myths, fears, stereotypes of and discrimination against disabled people continues. There is no free choice if women are under social pressure to accept routine tests. There can be no real choice until women feel able to continue with a pregnancy knowing that they will be bringing their child into a welcoming society that provides comprehensive systems of support (p. 6) [underline added for emphasis].

DPI's rhetorical construction centers prenatal testing as a conversation about autonomy more than about making particular reproductive choices. They frame their argument to question prospective parents' freedom to choose to accept or reject genetic testing independently of any decisions about pregnancy management. DPI indirectly acknowledges the evidence suggesting that knowledge about a particular trait itself does change prospective parents' desire for their future baby, with between 92% and 58% of prospective parents terminating a pregnancy following a definitive diagnosis of Down syndrome and Klinefelter syndrome, respectively

(Mansfield, Hopfer, & Marteau, 1999). The implicit question DPI raises is whether such information should have such an impact and why.

DPI critiques multiple elements of informed consent in prenatal genetic testing throughout their policy statement. They argue that: first, most clinicians have a limited understanding about living with disability (pp. 7,17); second, stereotypes about disability further limit prospective parents' understanding about whether or not to accept prenatal testing (pp. 6,8); third, social pressure to accept routine prenatal testing is coercive (pp. 6,7,10,12); fourth, disability discrimination may make prospective parents feel unable to knowingly continue pregnancy and feel ill-equipped to raise a disabled child (pp. 7,9). This rhetorical shift from informed consent to informed choice may be a useful strategy; the language highlights the importance of informed consent about testing, but also the possibility that this decision might lead to a choice for or against abortion. In this manner, DPI emphasizes that prospective parents need to have complete information about living with a disability and disability discrimination before making decisions about prenatal testing and pregnancy management following prenatal diagnosis.

The ACOG news releases do not explicitly use the word "choice". Nonetheless, they are explicit in their description of the practice of fetal chromosomal abnormalities screening as not required or even a recommendation. To illustrate this, they quote Dr. Driscoll, the primary author of the Practice Bulletin and an obstetrician-gynecologist (who also has certifications in medical genetics):

> We are not recommending that all pregnant women be screened but rather we are recommending that all pregnant women be *offered* screening. Physicians are ethically obligated to fully inform our patients of their healthcare options, including prenatal testing. It is entirely up to the patient whether or not she wishes to be screened for fetal chromosomal abnormalities without judgment from their physician [italics in original text].

ACOG asserts that prenatal testing involves decision-making on the part of each individual patient. Similarly to DPI, ACOG explicitly recognizes an extension of choice beyond that of the abortion debate, stipulating that their change in the practice guidelines affords women a greater range of options. In this manner, this medical community draws upon values about healthcare consumerism, where choice implies the consumer's ability to choose whichever tests from a range of healthcare services.

DPI may have reservations about the offer of prenatal testing that ACOG proposes, especially given the expressed pressures that they believe accompany it. As noted, DPI expressed concerns about the broader social context within which these decisions are made. DPI actively engages the language of choice. Arguably, this language use is deliberate, and it reminds the reader that these are choices, albeit constrained and sometimes misinformed ones. In contrast, ACOG successfully evades language of choice, and instead, ACOG makes reference to informed consent, acknowledging at least some of the bioethical issues at-stake. In doing so, attention focuses on the individual and her wishes, overlooking any social influences that may be at-play.

Murky Metaphors

DPI uses explicit metaphors connecting genetics to eugenics and to mass destruction. The repetitive use of "genetic" in conjunction with strategically placed references to elimination, manipulation, and human rights violations blurs any preconceived notion of genetics as distinct from eugenics. While the organization makes no attempt to define eugenics, they capture the underpinning values of eugenics. In particular, they try to take the negative association with eugenics and import it into the conversation about such testing. DPI only uses the word "eugenics" twice (pp. 3, 5), but these more subtle rhetorical connections are arguably more effective:

> Human genetics poses a threat to us because while cures and palliatives are promised, what is actually being offered are genetic tests for characteristics perceived as undesirable. This is not about treating illness or impairment but about eliminating or manipulating foetuses which may not be acceptable for a variety of reasons. These threaten our human rights (p. 3).

DPI suggests that if the purpose of prenatal screening is to eliminate particular fetuses, then the practice of such screening sends the message that the lives of people with disabilities are not worth living; they imply that our human rights are threatened when diversity is neither valued nor tolerated. They highlight that genetic testing provides a mechanism to identify differences that they suggest may be sought to be eliminated: "What we do oppose is genetic cleansing, driven by profit motive and social efficiency, informed by prejudice against disabled people and carried out in the name of cure or treatment." (p. 5). DPI conveys a variant of ethnic cleansing, where disability is analogous to ethnicity. Of particular significance is the notion that a powerful social group tries to eliminate another less powerful group; however, they strategically avoid explicit reference to the actors.

Given their consistent use of first-person, plural pronouns the audience may identify with the victim and can substitute whatever social player (i.e., biomedicine, economics, politics) as the agent with power. This rhetorical move is different from traditional metaphors of pro-life advocates who liken individual doctors to Nazi war criminals (Smith, 1995). The ambiguous actors leave interpretation to the audience, which may be a useful strategy; from within this rich discursive space, most people can identify a powerful actor and an oppressed one. DPI's framing of genetic testing focuses on its purpose and the social factors that might influence decision-making; whereas, abortion rhetoric often frames individual women or doctors in a negative light.

DPI also makes a direct association to nuclear power stating, "Nuclear energy is a source of life and a cause of death. If given an opportunity to express their opinion surely the victims of Nagasaki or Chernobyl would have fought for stricter regulation of the practical use of that new scientific knowledge. The same is true of the revolutionary developments in human genetics" (p. 3). While DPI makes explicit connections to the atomic bombings of Hiroshima and Nagasaki during World War II, DPI leaves some ambiguity and does not define either the victims or the agents with control of the genetics. As such, DPI creates a recurrent theme for the need of social responsibility in science that again situates their concern on the social dimensions that may make genetics problematic, as opposed to individual actors or individual choices.

ACOG does not adopt any of DPI's metaphors, and they also do not take the opportunity to acknowledge any social factors that may make genetic testing or the offer of such testing problematic. Instead, this medical community uses implicit or unintended metaphors relating to screening. Likely trying to differentiate between tests that identify risk for a particular condition (i.e., screening) from tests that identify specific genetic traits, ACOG avoids using the term "genetic testing", and instead, they consistently use "screening":

> According to the new guidelines, the goal is to offer screening tests with high detection rates and low false positive rates that also provide patients with diagnostic testing options if the screening test indicates that the patient is at an increased risk for having a child with Down syndrome (2007c) [underlines added for emphasis].

Perhaps unintentionally, the medical communities fail to recognize the metaphors at play when using the term screening. At its core, screening is about identifying and examining particular characteristics, but in definition, the purposes have evolved. While ACOG accurately compares screening from diagnostic tests, they fail to acknowledge that historically, the purpose of screening was to separate out and remove the unwanted (like with gold or coal). Interestingly,

the idea of separating out and removing the unwanted is exactly the message DPI suggests is morally questionable in relationship to prenatal tests.

According to Morabia & Zhang (2004), the United States Army implemented one of the earliest screening programs in 1917, using paper-based tests to identify potential recruits with psychiatric conditions as unfit for service. Nowadays, screening has become ubiquitous with healthcare; most people understand the purpose is to identify health conditions in people without signs or symptoms with the hopes of early treatment and management. Screening procedures − regardless of the condition of interest − focus on secondary prevention, which is early detection and when possible, treatment of pre-clinical manifestations (The Association of Faculties of Medicine of Canada Public Health Educators' Network).

This medical community draws upon what have become familiar discourses related to cervical cancer (e.g., a Pap smear) and/or sexually-transmitted infection screenings (e.g., HIV testing), and their words reflect this dominant understanding:

> ACOG revised its guidelines that now recommend offering fetal chromosomal screening to all pregnant women, regardless of age, because of improvements in low-risk, noninvasive screening methods" (2007a). Where facilities exist for additional screening methods, such as maternal serum screening, estimation of risk based on maternal age in isolation may not be appropriate [underlines added for emphasis].

ACOG's use of "screening to all pregnant women" could be viewed as an appeal to justice. This may be a useful rhetorical strategy, as the appeal to this distinct moral principle relates to different issues (i.e., autonomy might best address the decision to have a genetic test, whereas justice might best address access to the test). More central to this issue is the public understanding of screening; in particular, what is the benefit of screening? With cancer screening, the purpose is to identify cancer early, so that intervention (e.g. chemotherapy or tumor removal) is possible; alternatively, with sexually-transmitted infection screening, the

purpose is to identify the condition to begin treatment. Even in the absence of available cures, preventing transmission (e.g. mother-to-child, partner-to-partner) is an understood benefit. Particularly in a public sphere, this medical community use of the term screening in relationship to prenatal genetic testing may be problematic. In the case of prenatal testing for Down syndrome, no cure or treatment exists for the developing fetus following prenatal diagnosis; therefore, the information gained from testing ultimately leaves prospective parents with a decision about whether or not to end the pregnancy. As such, screening and prevention in this arena might suggest eliminating "undesirable" traits and preventing the birth of those who might otherwise live with such traits.

Missing Link

At present, there is no cure or treatment for any prenatal genetic diagnosis. The most recognizable benefit gained from testing is knowledge. Knowledge gained can reassure prospective parents about a "healthy" future child, give time to prepare for a child with a disability, or opportunity to interrupt the pregnancy. The medical community likely does not want to convey the message that pregnancy termination is even a potential benefit, especially to a public audience. As a profession, obstetricians and gynecologists are intimately involved with family planning and pregnancy management, including pregnancy termination. Given the contentious political-legal environment, practice guidelines may be reluctant to address abortion. Some health professionals might fear that such attention may ultimately constrain access to abortion services, as demonstrated by the fact that ACOG circumvents any discussion about pregnancy termination and makes no reference to abortion in these News Releases. Thus, this medical community presents screening for fetal chromosomal abnormalities as quite separate from pregnancy termination. Excluding language about abortion and reproductive choice may be a deliberate attempt to emphasize that this policy is about recognizing the value of

information for patients to make informed personal choices – not simply informed healthcare ones. At the same time, however, a clearer policy communication might state this upfront.

Especially given the purposes of a News Release that is intended for a broad public audience with diverse beliefs and values held about both prenatal testing and abortion, ACOG's representation is likely appropriate. However, if the potential relationship between prenatal testing and abortion is not conveyed in the public discourse, then the public is largely dependent upon patient-clinician interactions to make this connection. This approach may not promote education about the practice of prenatal testing to the public. As Rapp (1996) discusses in relationship to "positive" genetic diagnoses using amniocentesis, some prospective parents may misinterpret or misunderstand this tacit connection between screening and pregnancy termination.

Not only is the word "abortion" unsaid, ACOG substitutes "20th week of pregnancy" to address legal implications associated with the offer. Given the time needed to obtain results from invasive testing, the 20th week of pregnancy encroaches on the beginning of many states' restrictions of pregnancy termination. For example, the original News Release highlights:

> The new ACOG guidelines recommend that all pregnant women consider less invasive screening options for assessing their risk for Down syndrome, a common disorder that is caused by an extra chromosome and can result in congenital heart defects and mental retardation. Screening for Down syndrome should occur before the 20th week of pregnancy." (2007c) [underline added for emphasis].

They use this tactic in the subsequent News Release, as well. While this may help to distance their guidelines from some viewers, they also seemingly decontextualize relevant facts about prenatal genetic testing. Specifically, they do not acknowledge that such testing can lead prospective parents towards a series of pregnancy decisions that may end in pregnancy termination. Press & Browner (1997) discuss how many pregnant women understand routine

prenatal care, including prenatal genetic screening, as beneficial. In fact, many believe that such care helps to ensure a healthy baby. Interestingly, however, most such testing only provides information about a developing fetus, rather than any potential cure or remedy. Press & Browner (1997) note that while some pregnant women find this information to be reassuring and even empowering, fewer (13%) acknowledge that it may also reveal a diagnosis of a condition, for which they may choose to terminate the pregnancy. Additionally, Press & Browner (1994) suggest that the topic of abortion is absent from patient education materials. Rather than obscuring the potential connection between such testing and pregnancy management decisions, ACOG should make greater efforts to develop and to promote health education materials related to prenatal testing; this would also help to address critiques about physicians failing to provide fully informed consent.

Not surprisingly, DPI references abortion. The repeated, strategic use of choice may implicitly bring to mind abortion rhetoric, and their use of the word of abortion may further substantiate the connection. Most of DPI's usage of the word "abortion" specifically refers to the current legal status of such services. For example, "We are threatened by abortion laws which discriminate against the birth of disabled children" (p. 4). While the policy statement does not provide any further explanation about the expressed discrimination in abortion laws, this might be in reference to legal arguments and/or laws that suggest that the offer of prenatal testing is problematic in instances where abortion is illegal, as prospective parents do not have to opportunity to take action on the information gained from the testing; in this manner, some suggest that at the very least in areas where prenatal testing is available, abortion on the basis of

disability should also be legally allowable.[8] Alternatively, DPI make be making reference to wrongful birth or wrongful life; these legal causes of action allow healthcare professionals and healthcare systems to be sued for failing to disclose a prenatally diagnosed condition, with the rationale that such affected-pregnancies would have been terminated in light of the information.[9] This explicit connection between abortion and law may be particularly problematic for those concerned about preserving the availability of legal abortions, especially if they share concern about discrimination on the basis of disability status. If DPI aims to promote further public discussion about whether or not the practice of prenatal testing should continue as is, then perhaps, selecting to use the word "abortion" in conjunction with "law" should be limited. Instead, DPI might still educate and inform the public about these issues by explaining how restricting access to abortion services can in fact have discriminate against the birth of children with disabilities, by making disability an unjustified exception in policy and by affirming negative biases and assumptions about disability in practice. This language use may surface underlying fears about altering the legal status of abortion laws, which may fail to appeal to an audience who is sympathetic to their cause. Furthermore, those reluctant to draw attention to politically susceptible abortion laws may not engage in public dialogue about the practice of prenatal testing.

---

[8] This is a contested debate worth acknowledging in this context; however, as it is somewhat tangential to the central analysis here, I simply direct interested readers to a collection of further reading (Aramesh, 2009; Ballantyne, Newson, Luna, & Ashcroft, 2009; Buccafurni & Chang, 2009; Kon, 2009; Sperling, 2009; Wasserman & Asch, 2009; Zivotofsky & Jotkowitz, 2009).

[9] While relevant to the discussion, the debate surrounding these is tangential to the central analysis here, and I simply direct readers to the legal precedent for wrongful birth ("Azzolino v. Dingfelder," 1985; "Gallagher v. Duke University," 1988; "Munro v. Regents of University of California," 1989) and wrongful life ("Harbeson v. Parke-Davis, Inc," 1983; "Procanik v. Cillo," 1985; "Turpin v. Sortini," 1982).

CONCLUSION

Given the ever-growing availability of genetic testing, including the application of cell-free fetal DNA analysis (Hahn & Holzgreve, 2002), this realm of public policy will become increasingly important to healthcare and public health. Committee opinions about noninvasive methods of prenatal genetic diagnosis have been recently published (Devers *et al.*, 2012; Langlois, Brock, & Genetics Committee, 2013; The Noninvasive Prenatal Screening Work Group, 2013), so it is foreseeable that medical communities will craft new practice guidelines within a short timeline. This creates an opportunity for disability rights organizations and other health advocates to draft policy statements addressing these new technologies. In order to effectively communicate their values and beliefs to a public audience, such organizations must make explicit and specific claims about what in current practice medical professionals are doing poorly; in other words, the feedback ought to be more constructive, so that there are clearer pathways to actionable changes. In an era of evidence-based medicine, these organizations would benefit from utilizing existing scholarship and creating a sound empirical foundation for their claims, rather than relying solely upon anecdotes and speculations. Collaborating with sympathetic and supportive health professionals may be key to success. As discussed, statements might also benefit from more direct critique of the offer and practice of prenatal testing, allowing for some distancing from ideologically grounded and contentious debates about abortion; that is not to say that abortion should be completely eliminated from a conversation, but rather that the focus should remain upon how we as a society ought to make reproductive choices less constrained and better informed. Even if such statements are completely disregarded by existing medical establishments, connecting with the general public may create opportunities to educate others about disability and to identify new allies who may also be wary

of mass implementation and widespread adoption of prenatal genetic testing or concerned about the potential erosion of informed medical choices. In so doing, disability communities ensure their representation as a stakeholder in this ongoing public issue.

Since the undertaking of the Human Genome Project (US Department of Energy Genome Programs, 2011), disability communities have been notable critics, while the medical communities have largely supported prenatal genetic testing. By directing the conversation to address specific issues at stake with prenatal genetic testing, disability communities can help to continue providing more balanced coverage of disability representations to the public and to guide clinical practice. However, many of the critiques of prenatal testing have come from disability-specific organizations. Given that emerging technological advances allow prenatal tests to identify a wider range of genetic conditions, I think that future recommendations should increasingly reflect cross-disability alliances. Coordinated, unified efforts − across disability communities and with allies − are needed to influence this public conversation. This analysis suggests that disability communities must delve into precisely how clinicians are misinforming or not providing informed consent, in order to highlight mechanisms to improve upon current practice. These critiques would be more effective, if they are grounded in empirical research that identifies what actually takes place within the clinical encounter, in order to provide the foundation for reflecting on how misinforming or incomplete informed consent might occur. For example, disability communities might want to identify specific issues about living with disability that are absent or poorly represented in health education materials. Alternatively, disability communities might provide greater clarity about how genetic counseling is directive, despite professional tenets that suggest value-neutrality and nondirective counseling. Additionally, disability communities have to make efforts to engage directly with prospective

parents who may need and want the information that the disability communities can provide. Publishing position statements on the Internet alone is insufficient; efforts must be made to enter mass media, including newspapers, health education materials, and radio. In order to make a public case for diversity, disability communities need to engage with local healthcare systems, media outlets, and beyond. By utilizing multiple mechanisms, disability communities can help to transform a private, patient-provider encounter into a public conversation, in order to enhance reproductive autonomy and to further promote disability as valued diversity.

Through examining policy statements, this analysis reveals how the deliberate word choices may also re-shape conversations about abortion. The disability and medical communities actively extend the notion of choice beyond that of abortion rhetoric. Disability communities transformed informed consent into informed choice, highlighting the importance of choice in this context, as well as the need for accurate, up-to-date information and structural supports, in order for prospective parents to make them freely. In contrast, medical communities focus the conversation on healthcare consumerism. Drawing upon this notion of healthcare consumerism, future policy statements from disability communities may want to make space to address other related, but equally relevant issues, like home- and community-based services, disability representations in health education materials, and education and employment opportunities. These issues can constrain prospective parents' understandings about the capability of a child with a disability, and they also build upon the notion that individuals should have a range of available options (e.g., not only the option to choose a child with a disability, but also the option to raise such a child in their own home and community; disability representations that show people with disabilities flourishing in their lives, rather than predominantly focused on

cures and treatments; people with disabilities who have equal access to a diverse set of education and employment opportunities as their peers without disabilities).

While the medical communities did not adopt this language of choice, some address the issue of informed consent. However, they do not readily address the relationship between prenatal genetic testing and pregnancy management. In this rhetorical move, they neglect the disability community's critique that a woman's choice to have or not have prenatal genetic testing should reflect what parents think is the purpose of testing. The use of metaphors about eugenics and screening creates multiple opportunities for the audience to engage with their argument, and given that the audience likely comes from diverse political and moral backgrounds, this technique may be a useful strategy. On the other side, medical communities − perhaps unintentionally, use the word screening − tapping into multiple meanings, which given their equally diverse audience, may pose problematic. The notion of screening with the purpose to eliminate builds upon the concerns of the disability communities. I would not say that the word screening is itself the problem, but rather that medical communities should be more transparent about the potential connection between such prenatal tests and pregnancy management choices. A more transparent policy communication might specify that the offer of screening reflects a commitment of the organization to provide women with valuable information, so that they are empowered to make informed personal and healthcare decisions. Furthermore, ACOG might include language stating that decisions about whether to test, which tests, and when, as well as whether to continue or end a pregnancy are individual patient's choices. While I recognize that medical communities may be reluctant to address abortion head-on in a practice guideline, its absence suggests that the information available during decision-making is vague; excluding abortion from the practice guideline fuels existing concerns about

public misunderstandings and constrained reproductive choices. Medicine is a model for other health professionals and possesses the power to influence policy, practice and patient education. Therefore, I would argue that medical communities have a responsibility to the public to be open about abortion in this context. While this may initiate unwanted public conversations about reproductive autonomy, I think it also presents an opportunity to ensure that practice guidelines reflect the need for providers to discuss with their patients that pregnancy termination is a chosen outcome for some patients.

By looking at the rhetoric of both the disability and medical communities together, we see that public conversations about prenatal genetic testing are notably complex. Both groups are similarly entangled in multiple discourses and perhaps unintentionally engaged in issues unrelated to the task of their policy statements. Thus, those trying to covey messages about prenatal genetic testing to the public need to be mindful not just of the science, but also of the rhetorical elements at play − particularly as the rhetoric provides the connection to values and assumptions that likely influence how the audience interprets what is being said. Careful use of language can help to navigate the competing discourses and to get at shared values, in order to answer fundamental questions: should prenatal genetic testing be offered, for which conditions, and if so, how?

References

ACOG Office of Communications. (2007a). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

ACOG Office of Communications. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm

Aramesh, K. (2009). A closer look at the abortion debate in Iran. *The American Journal of Bioethics, 9*(8), 57-58.

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.

Asch, A., & Wasserman, D. (2005). Where Is the Sin in Synecdoche? Prenatal Testing and the Parent-Child Relationship. In D. T. Wasserman, R. S. Wachbroit & J. E. Bickenbach (Eds.), *Quality of life and human difference : genetic testing, health care, and disability*. New York: Cambridge University Press.

Azzolino v. Dingfelder, No. No. 718PA84, 337 528 (NC: Supreme Court 1985).

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? *The American Journal of Bioethics, 9*(8), 48-56.

Beauchamp, T. L., & Childress, J. F. (1994). *Principles of biomedical ethics*. New York: Oxford University Press.

Bérubé, M. (1998). *Life as we know it : a father, a family, and an exceptional child*. New York: Vintage Books.

Buccafurni, D., & Chang, P. L. (2009). Does prenatal diagnosis morally require provision of selective abortion? *The American Journal of Bioethics, 9*(8), 65-67.

Condit, C. M. (1990). *Decoding abortion rhetoric : communicating social change*. Urbana: University of Illinois Press.

Davis, D. S. (2010). *Genetic dilemmas : reproductive technology, parental choices, and children's futures*. Oxford; New York: Oxford University Press.

Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2012). Noninvasive prenatal testing/noninvasive prenatal diagnosis: The position of the national society of genetic counselors. *Journal of genetic counseling*, 1-5.

Disabled People's International. (2000). Disabled Persons Speak on the New Genetics.

Disabled People's International. (2012). from http://www.dpi.org/

Eddleman, K. A., Malone, F. D., Sullivan, L., Dukes, K., Berkowitz, R. L., Kharbutli, Y., . . . D'Alton, M. E. (2006). Pregnancy loss rates after midtrimester amniocentesis. *Obstetrics and gynecology, 108*(5), 1067-1072.

Gallagher v. Duke University, No. No. 87-1056, 852 773 (Court of Appeals, 4th Circuit 1988).

Gee, J. P. (2010). *An introduction to discourse analysis : theory and method*. New York [etc.]: Routledge.

Ginsburg, F., & Rapp, R. (2010). Enabling Disability: Rewriting Kinship, Reimagining Citizenship. In L. J. Davis (Ed.), *The disability studies reader* (3rd ed., pp. xviii, 451 p.). New York: Routledge.

Hahn, S., & Holzgreve, W. (2002). Prenatal diagnosis using fetal cells and cell-free fetal DNA in maternal blood: what is currently feasible? *Clinical Obstetrics and Gynecology, 45*(3), 649.

Harbeson v. Parke-Davis, Inc, No. No. 48331-1, 656 483 (Wash: Supreme Court 1983).
Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. *Qual Health Res, 15*(9), 1277-1288. doi: 10.1177/1049732305276687
Kon, A. A. (2009). Prenatal testing can be advantageous even when abortion is not an option. *The American Journal of Bioethics, 9*(8), 69-70.
Langlois, S., Brock, J.-A., & Genetics Committee. (2013). Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. *J Obstet Gynaecol Can, 35*(2), 177-181.
Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. *American Journal of Human Genetics, 51*(4), 936.
Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. *The American Journal of Human Genetics, 62*(4), 768-775.
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. *Prenatal diagnosis, 19*(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b
Morabia, A., & Zhang, F. (2004). History of medical screening: from concepts to action. *Postgraduate medical journal, 80*(946), 463-469.
Munro v. Regents of University of California, No. Docket No. B037779, 215 977 (Cal: Court of Appeal, 2nd Appellate Dist., 7th Div. 1989).
Nadler, H. L. (1968). Antenatal detection of hereditary disorders. *Pediatrics, 42*(6), 912-918.
Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. *Health Expectations, 11*(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x
Press, N., & Browner, C. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.
Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.
Procanik v. Cillo, 502 94 (NJ: Superior Court, Law Div. 1985).
Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), *Representations of motherhood*: Yale University Press.
Roe v. Wade, 410 113 (Supreme Court 1973).
Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), *The disability studies reader* (2nd ed., pp. xviii, 451 p.). New York: Routledge.
Seavilleklein, V. (2009). Challenging the rhetoric of choice in prenatal screening. *Bioethics, 23*(1), 68-77.
Smith, L. (1995). Antiabortion group's leaflets liken doctors to war criminals. *Washington Post,* p. 3.
Sperling, D. (2009). From Iran to Latin America: must prenatal diagnosis necessarily be provided with abortion for congenital abnormalities? *The American Journal of Bioethics, 9*(8), 61-63.
The Association of Faculties of Medicine of Canada Public Health Educators' Network. Basic concepts in prevention, surveillance and health promotion. In D. Donovan, B. McDowell

& D. Hunter (Eds.), Primer on Population Health: The Association of the Faculties of Medicine of Canada. Retrieved from http://phprimer.afmc.ca/Part1-TheoryThinkingAboutHealth/Chapter4BasicConceptsInPreventionSurveillanceAndHealthPromotion/Thestagesofprevention.

The Noninvasive Prenatal Screening Work Group. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. *Genetics in Medicine, 15*(5), 395-398.

Turpin v. Sortini, No. Docket No. SF 24319, 643 954 (Cal: Supreme Court 1982).

US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml

Varvasovszky, Z., & Brugha, R. (2000). How to do (or not to do) . . . A stakeholder analysis. *Health policy and planning, 15*(3), 338-345.

Wasserman, D., & Asch, A. (2009). An unjustified exception to an unjust law? *The American Journal of Bioethics, 9*(8), 63-65.

Wilkinson, S. (2008). "Eugenics talk" and the language of bioethics. *Journal of Medical Ethics, 34*(6), 467-471. doi: 10.1136/jme.2007.021592

Zivotofsky, A. Z., & Jotkowitz, A. (2009). Prenatal diagnosis and abortion are not in conflict in Israel. *The American Journal of Bioethics, 9*(8), 58-60.

With increasing amounts of information available online, health education is often received outside the clinical encounter. As such, information-seeking prospective parents may engage with health education about prenatal testing before pregnancy, before prenatal screening, after prenatal screening results, before choosing prenatal diagnostic testing, or after diagnostic results. In this section, I explore themes in a purposive sample of health education materials, including both online and printed ones. This analysis focuses on how these materials create and shape reproductive choices that patients may consider.

CHAPTER 3 HEALTH EDUCATION

In their foundational work "Collective silences, collective fictions", Press & Browner (1994) examined health education brochures sponsored by the State of California and offered to prospective parents about maternal serum alpha fetoprotein (AFP) screening. They articulate the collective fiction is the simple, routine offer of such testing that results from healthcare providers' discomfort related to discussing selective termination and patients feel contemplating it; this fiction creates a collective silence from patients and providers about the potential outcomes that may arise following such testing. They identified several notable issues in relationship to patient understanding following distribution of such materials. First, they found that these health education brochures at no instance described the positive test result depicted as a "negative" event. From a health education perspective, some might argue that this is a good portrayal, given that no value or meaning is attached to any particular result. As Press & Browner suggest, however, such representation may inadequately inform prospective parents about the potential outcomes of a testing result. Second, Press & Browner found that there was no mention of the possible, chosen outcome of pregnancy termination following a positive diagnosis. Third, upon assessing women's understanding of AFP after receiving this brochure, they also discovered that only 75% read it and felt they understood it (Press & Browner, 1994). Many prospective mothers understood prenatal genetic testing as a means of reassuring themselves and their physicians about their developing fetus' health (Press & Browner, 1997).

Press and Browner's work directs attention to the clinical encounter, identifying potential barriers to informed consent; in particular, this work highlights the need for patients to have both the information necessary to make a decision about testing and sufficient understanding to enact a decision reflective of their values. More recent work continues to highlight that patients may have inadequate knowledge about prenatal testing for Down syndrome (Kuppermann *et al.*, 2015; van den Berg, Timmermans, ten Kate, van Vugt, & van der Wal, 2006), receive imbalanced or bias messages about Down syndrome (Bakke, 2013; Kellogg, Slattery, Hudgins, & Ormond, 2014; Lawson, Carlson, & Shynkaruk, 2012), feel pressured to accept routine tests (Lewis, Silcock, & Chitty, 2013; Stapleton, Kirkham, & Thomas, 2002), and/or evaluate and use different knowledge sources that do not lend themselves to a straightforward conclusion and testing decision (Markens, Browner, & Preloran, 2010; Potter *et al.*, 2008).

In recent years, the health education landscape is dramatically changing – shifting from predominately paper brochures received within the clinic to a wealth of online and electronic resources publically available through causal web browsing. In fact, according to Pew Research Center, 80% of adult Internet users in the United States look for health information online, and over one-third have read about someone else's commentary or experience of a medical issue or treatment online (Fox, 2012). While there is little, if any, scholarly work tackling why information-seeking behaviors are evolving, I speculate that there is: simply more information available than previously; questions can be asked privately −or at least seemingly so− without feeling threatened or foolish in front of a medical provider; the information is freely available to us and we can conveniently access this information in a manner that fits our schedules as opposed to a clinical visit; accessing information online ensures that we have access to more, potentially diverse perspectives.

As such, prospective parents may engage with information about prenatal testing at multiple time points and access points, as well as from an array of sources and locations. There is some evidence suggesting that the public trusts traditional print media sources more so than online ones (Media Insight Project *et al.*, 2014). However, to the best of our knowledge, to date, there is no investigation of how online health education materials compare to print ones with regards to their trustworthiness or content quality. In efforts to better understand what information is available to prospective parents, this study evaluates online and print health communication materials about prenatal genetic testing for Down syndrome distributed by nonprofit and government organizations, as well as primary care and genetic specialty clinics. The point is not to suggest that online or print material is better than the other, but rather to offer ways in which print and online materials might complement each other, in order to improve patient comprehension and to empower decision-making consistent with patient values.

METHODS

By obtaining health education materials directly from practitioners, I identified health education materials developed and distributed from multiple disciplinary perspectives, including public health, genetic counseling, obstetricians and gynecologists. I also collected online, publically searchable health education materials. In total, a purposive sample of 30 health education materials was analyzed (Table 3). Paper materials were scanned, and digital versions were downloaded. All texts were imported into Atlas.ti 7 ™ for analysis.

Using a set of guiding questions (Table 4), the analysis employed a form of directed content analysis (Hsieh & Shannon, 2005). Each text was evaluated based upon a set of *a priori* criteria, based upon different issues raised in the literature: ease of access (Moorhead *et al.*, 2013); accurate and sufficient information provided about related concepts about risk

communication, genetic education and labeling and language use (Reed, 2009); options and potential outcomes explained (Press & Browner, 1994); potential financial implications and conflicts of interest disclosed; diverse patient supports offered. The health education materials were qualitatively explored to address broad concepts, including the purposes of testing, the benefits, harms, and costs; the framing of disability; as well as key actors and their potential conflicts of interest. Through an iterative process, two independent reviewers coded these texts. When discrepancies arose, both independent coders returned to the original full-text together and discussed the article passages that led them to their classification; through conversation about relevant passages, they reconciled these discrepancies and came to consensus about their assessment of key passages. The codes were built into broad themes.

Table 3. Included health education materials

Number	Title	Discipline	Distribution	Region
1	Frequently asked questions about genetic disorders	OB/GYN	Electronic	National
2	Frequently asked questions about routine tests in pregnancy	OB/GYN	Electronic	National
3	Frequently asked questions about screening for birth defects	OB/GYN	Electronic	National
4	Quick tips for finding, evaluating, processing, and organizing information on the web	OB/GYN	Electronic	National
5	Prenatal care	Public Health	Electronic	National
6	Birth defects	Public Health	Electronic	National
7	Quad Screen	Clinical	Electronic	National
8	Understanding a diagnosis of Down syndrome	Advocacy organization	Electronic	National
9	National Down Syndrome Society	Advocacy organization	Electronic	National
10	A promising future together: A guide for new and expectant parents	Advocacy organization	Electronic	National
11	What is Down syndrome?	Advocacy organization	Electronic	National
12	Amniocentesis	Government organization	Electronic	National
13	Understanding Down syndrome diagnosis	Advocacy organization	Electronic	National
14	First trimester Down syndrome screen	Public and private, electronic medical record system	Electronic	National
15	Prenatal services	Private hospital / Genetic counseling	Electronic	Local
16	Guide to prenatal testing	Non-profit hospital / Genetic counseling	Brochure	Local
17	Advanced aneuploidy screening	Non-profit hospital / Genetic counseling	Brochure	Local

Number	Title	Discipline	Distribution	Region
18	Chorionic villus sampling	Non-profit hospital / Genetic counseling	Brochure	Local
19	Integrated screen	Non-profit hospital / Genetic counseling	Brochure	Local
20	Quad screen	Non-profit hospital / Genetic counseling	Brochure	Local
21	Down syndrome	Private hospital / Genetic counseling	Electronic	Local
22	Frequently asked questions for Harmony testing	Private hospital / Genetic counseling	Electronic	Local
23	Down syndrome	Advocacy organization	Electronic	National
24	Prenatal health	Private hospital / OB/GYN	Brochure – Corporate	Local
25	First screen	Private hospital / OB/GYN	Brochure – Corporate	Local
26	Integrated screen	Private hospital / OB/GYN	Brochure – Corporate	Local
27	Facts about Down syndrome	Government / Public Health	Electronic	National
28	Diagnosis	Government / Public Health	Electronic	National
29	Harmony prenatal test	Private hospital / OB/GYN	Brochure – Corporate	Local
30	Maternal screening an important part of prenatal care	Private hospital / Laboratory Services	Brochure – Corporate	Local

Table 4. Health education analysis guide

Criteria	Explanation
Ease of access	• What level of effort would be required for the average member of the public to obtain access to this material? • What, if any, barriers exist to accessing material? • What, if anything, facilitates access to this material?
Accurate and sufficient information provided about related concepts	Risk communication • Are concepts related to risk explained in an understandable way? • Are risk factors described? If so, how? • Are particular subgroups identified as at-risk? Genetics education • Are concepts related to genetics adequately explained for the general public? • Is the genetics of Down syndrome described? If so, how? • What details are provided or left out? Is this reasonable compared to other sources? Labeling and language use • Is any potentially problematic or stigmatizing language used? If so, what? • Are any groups labelled differently or framed in a potentially negative light? • What, if any, further information is provided to explain or destigmatize particular groups?
Accurate and sufficient information provided about testing methods	• Are different testing methods discussed, as appropriate? • Are screening and diagnostic testing methods differentiated and explained? • Are the purpose(s) discussed? • Does it mention that having such testing is voluntary or a personal choice?
Options and potential outcomes explained	• Are potential benefits mentioned? • Are potential harms mentioned? • Is it mentioned that information gained may impact future pregnancy management decisions?
Potential financial implications and conflicts of interest disclosed	• Are financial costs discussed? • Who authored the material? • Does any commercial interest exist? If so, is this disclosed?
Diverse patient supports offered	• Are readers directed to healthcare professionals or health professions organizations for further questions or information?

Criteria	Explanation
	• Are readers provided with contact information or guidance towards nonclinical support and resources? • Are readers offered sources to double check the accuracy, completeness or the evidence to support the information offered?

Table 5. Illustrative examples

Criteria	Explanation
Accurate and sufficient information provided about related concepts	Risk communication "The risk of having a child with a chromosomal disorder increases as a woman ages. For instance, a 35-year-old woman has a 1 in 192 (less than 1%) chance of having a baby with a chromosomal disorder. The chance increases to 1 in 66 (about 1.5%) in a woman aged 40 years." (1) "In general, in each subsequent pregnancy the chance of having another baby with Down syndrome is about 1 in 100 up to age 40. After age 40, the risk is based on the mother's age. If, however, the first child has translocation Down syndrome, the chance of having another child with Down syndrome may be greatly increased." (6) "Results of the quad screen indicate your risk of carrying a baby who has certain chromosomal conditions, such as Down syndrome." (7) "Once a woman has given birth to a baby with Trisomy 21, it is estimated that her chances of having another baby with Trisomy 21 is 1% greater than her chances by age alone." (9) "Maternal age, however, is not linked to the chance of having a baby with translocation. Most cases are sporadic, chance events, but in about one third of translocation cases one parent is a carrier of a translocated chromosome." (10) "There is no definitive scientific research that indicates that Down syndrome is caused by environmental factors or the parents' activities before or during pregnancy." (11) "The older a pregnant mother is, the higher her chance is of having a baby with a genetic disease." (12) "The risk for chromosomal abnormalities gradually increases with age, but a woman of any age can have a baby with a chromosomal abnormality." (15) "The mother's age at delivery is the only factor found to be linked to the risk of having a baby with Down syndrome." (21)

Criteria	Explanation
	"While you can decrease the incidence of some risk factors, others are not in your control. Also, not all risk factors will have an effect on outcomes in all pregnant women and babies." (24) "The risk of having a child with Down syndrome gradually increases with the age of the mother, but can occur at any maternal age." (25) "One factor that increases the risk for having a baby with Down syndrome is the mother's age." (27) "Women age 35 and older have an increased chance of giving birth to a baby with Down syndrome." (30) Genetics education "Down syndrome is a caused by extra genetic material from chromosome 21. Chromosomes are the structures in cells that contain the genes. Each person normally has 23 pairs of chromosomes, or 46 in all. An individual inherits one chromosome per pair from the mother's egg and one from the father's sperm. When an egg and sperm join together, they normally form a fertilized egg with 46 chromosome. Sometimes something goes wrong before fertilization. A developing egg or sperm cell may divide incorrectly, sometimes causing an egg or sperm to have an extra chromosome 21. When this cell joins with a normal egg or sperm cell, the resulting embryo has 47 chromosomes instead of 46. Down syndrome is called trisomy 21 because affected individuals have three number 21 chromosomes instead of two. This type of error in cell division causes about 95 percent of the cases of Down syndrome. Occasionally, before fertilization, a part of chromosome 21 breaks off during cell division and becomes attached to another chromosome in the egg or sperm cell. The resulting embryo may have what is called translocation Down syndrome. Affected individuals have two normal copies of chromosome 21 plus extra chromosome 21 material attached to another chromosome. This type of error causes about 3-4 percent of the cases of Down syndrome. In some cases the parent has a rearrangement of chromosome 21 called a balanced translocation, which does not affect his or her health. About 1-2 percent of individuals with Down syndrome have a form called mosaicism. In this form the error occurs after fertilization. Affected individuals have some cells with an extra chromosome 21 and others with a normal number." (6/7)

Criteria	Explanation
	"Down syndrome is usually caused by an error in cell division called 'nondisjunction'. Nondisjunction results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of the 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This type of Down syndrome, which accounts for 95% of cases is called Trisomy 21. The two other types of Down syndrome called mosaicism and translocation. Mosaicism occurs when nondisjunction of chromosome 21 takes place in one but not all-of the initial cell divisions after fertilization. When this occurs, there is a mixture of two types of cells, some containing the usual 46 chromosomes and others containing 47. Those cells with 47 chromosomes contain an extra chromosome 21. Mosaicism accounts for about 1% of all cases of Down syndrome. Research has indicated that individuals with mosaic Down syndrome may have fewer characteristics of Down syndrome than those with other types of Down syndrome. However, broad generalizations are not possible to the wide range of abilities people with Down syndrome. Translocation accounts for about 4% of all cases of Down syndrome. In translocation, part of chromosome 21 breaks off during cell division and attaches to another chromosome, typically chromosome 14. While the total number of chromosomes in the cells remain 46, the presence of an extra part of chromosome 21 causes the characteristics of Down syndrome. Regardless of the type of Down syndrome a person may have, all people with Down syndrome have an extra, critical portion of chromosome 21 present in all or some of their cells. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome." (9/10/11) "Down syndrome is a condition that is usually caused by an extra copy of the 21st chromosome-genetic building blocks of all people." (13) "Aneuploidy is when a person has extra copies are missing copies of certain chromosomes. Most people have to copies of each of 23 different chromosomes, for a total of 46. Trisomy is when there are three copies of a certain chromosome in all the cells in the body." (17) "Normally in reproduction, the egg cell of the mother and the sperm cell of the father start out with the usual number of 46 chromosomes. The egg and sperm cells both undergo a cell division in which the 46 chromosomes are divided in half, so that both the egg and the sperm cells will have 23 chromosomes each. When a sperm with 23 chromosomes fertilizes an egg with 23 chromosomes, the baby will have a complete set of 46 chromosomes, half from the father and half from the mother. Sometimes, an error

Criteria	Explanation
	occurs when the 46 chromosomes are being divided in half, and an egg or sperm cell keeps both copies of the 21st chromosome instead of just one copy. If this egg or sperm is fertilized, and the baby will have three copies of the #21 chromosome, which is called trisomy 21, or Down syndrome. The features of Down syndrome are caused by the extra copy of chromosome #21 being in every cell in the body. Most cases of Down syndrome are caused by trisomy 21. Occasionally, the extra chromosome #21 or a portion of it is attached to another chromosome in the egg or sperm; this may cause 'translocation Down syndrome'. This is the only form of Down syndrome that may be inherited from a parent. Some parents have a rearrangement called a balanced translocation, in which the #21 chromosome is attached to another chromosome, but it does not affect their own health. Rarely, another form called 'mosaic Down syndrome' may occur when an error in cell division happens after fertilization. These individuals have some cells with an extra chromosome #21 (47 chromosomes total), and other cells have the usual number (46 total)." (21) "Down syndrome is a condition in which a person has an extra chromosome. Chromosomes are small 'packages' of gene in the body. They determine how a baby's body forms during pregnancy and how the baby's body functions as it grows in the womb and after birth. Typically, a baby is born with 46 chromosomes. Babies with Down syndrome have an extra copy of one of these chromosome, chromosome 21. A medical term for having an extra copy of a chromosome is 'trisomy'. Down syndrome is referred to as Trisomy 21. This extra copy changes how the baby's body and brain develop, which can cause both mental and physical challenges for the baby. (27) Labeling and language use "Chromosomal disorders are disorders caused by missing, damaged, or extra chromosomes. These problems often are caused by an error that occurred when the egg or sperm was forming. Most children with chromosomal disorders have physical defects, and some have mental defects." (1) "Down syndrome – mental retardation, abnormal features of the face, and medical problems such as heart defects occur as a result of an extra chromosome 21 (trisomy 21)." (3) "Down syndrome is a chromosomal disorder that includes a combination of birth defects. Affected individuals have some degree of intelligential disability, characteristics facial feature and, often, heart

Criteria	Explanation
	defects and other health problems. The severity of these problems varies greatly among affected individuals." (6) "A few of the common physical trait of Down syndrome are low muscle tone, small stature, an upward slant of the eyes, and a single deep crease across the center of the palm – although each person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all." (11) "An example of a genetic disease due to an abnormal chromosome is Down syndrome. Most people have two #21 chromosomes but people with Down syndrome have three 21's. People with Down syndrome have mental and physical retardation." (12) "The range of medical conditions and abilities can vary widely for people with Down syndrome. Each person with Down syndrome has his or her own strength and weakness that no one can predict before birth. People with Down syndrome have mild to moderate intellectual disabilities, low muscle tone, and higher chance of health issues, particularly heart conditions. While people with Down syndrome do face challenges, recent advances in healthcare, education and attitudes have greatly improved their lives. This progress has given them more opportunities as valued members of their communities. This means that more people with Down syndrome are finishing school, finding jobs, and having relationships." (13) "Down syndrome is also known as trisomy 21. It is caused when a person has an extra copy of chromosome number 21. Down syndrome affects people in different ways. People with Down syndrome always look different than other members of their family. They always have some developmental delay, but the level of delay differs from person to person. Adults with Down syndrome may be able to play sports, have a basic job, and enjoy friends. But they usually cannot live on their own without help. Many babies with Down syndrome have a heart defect, which can sometimes be fixed with surgery. Other health problems and birth defects sometimes occur with Down syndrome, but they are rare." (16) "Trisomy 21 is when there are three copies of the chromosome number 21 in all cells. It is the most common cause of a genetic condition called *Down syndrome*." (17)

Criteria	Explanation
	"Down syndrome is a genetic disorder that involves birth defects, intellectual disabilities, characteristic facial features. Additionally, it often involves heart defects, visual and hearing impairments, and other health problems. The severity of all these problems varies greatly amount affected individuals." (21) "Down syndrome is a chromosome condition associated with intellectual disability, a characteristic facial appearance, and low muscle tone in infancy. The degree of intellectual varies from mild to severe. People with Down syndrome may be born with a variety of health concerns, including heart defects or digestive abnormalities. In addition they have greater risk of developing gastroesophageal acid reflex, celiac disease, hyperthyroidism, vision problems, leukemia and Alzheimer's disease." (23) "Even though people with Down syndrome might act and look similar, each person has different qualities. People with Down syndrome usually have an IQ "a measure of intelligence" in the mild or moderately low range and are slower to speak than other children." (27) "Trisomy 21 called Down syndrome, is associated with mild to moderate intellectual disabilities and may also lead to digestive disease and congenital heart defects." (29)
Accurate and sufficient information provided about testing methods	• Are screening and diagnostic testing methods differentiated and explained? "If the results of the screening test or other factors raise concerns about your pregnancy, diagnostic tests can be done to provide more information." (3)" "Diagnostic tests can provide a definitive diagnosis with almost 100% accuracy." (11) "If the results of the screening test than normal, doctors usually offer further diagnostic testing to determine if birth defects or other possible problems with the baby are present. These diagnostic tests are offered to women with higher risk pregnancies, which may include women who are 35 years of age or older; women who had a previous pregnancy affected by a the birth defect; women who have chronic diseases such as this high blood pressure, diabetes, or epilepsy; or women who use certain medications." (6)

Criteria	Explanation
	• Are the purpose(s) discussed? "Diagnostic tests detect whether certain birth defects are present." (1) "Amniocentesis (also called amnio) is a common prenatal test used to diagnose certain birth defects and genetic conditions. Genetic conditions are health conditions and birth defects that are passed down to a baby from mom and dad. They may cause health problems the baby." (5) "Prenatal diagnostic procedures are specialized tests that can accurately diagnose certain birth during pregnancy." (15) • Does it mention that having such testing is voluntary or a personal choice? "Whether or not to undergo a prenatal screening and diagnostic test is a personal decision, and expectant parents must make the choice that is best for them." (13) "Choosing whether to have any of these test, or deciding which ones are best for you, can be hard. There is no "right" choice. Some women choose only an anatomy ultrasound and no other tests. Others may choose an integrated screen and anatomy ultrasound. And, if one of these tests is abnormal, they may have amniocentesis. Some women prefer a CVS or amniocentesis without any of the screening tests." (16) "This handout gives information to help you decide if you want to have an advanced aneuploidy screening test. Having this test is up to you. Some people do not find this type of test to be helpful. You may refuse testing at any time." (17) "Having this test is up to you." (19) "Doing a quad screen is your choice, and the decision is a personal one." (20)

Criteria	Explanation
	"When a woman finds she is pregnant, she faces many choices. One important choice is whether to have a maternal serum screening test, such as FirstScreen, to determine if she is at increased risk of having a baby with certain birth defects" (25) "The decision to consent to, or to refuse the above test is entirely mine." (29)
Options and potential outcomes explained	• Are potential benefits mentioned? "A prenatal screening tests named 'MaternT21,' has been made available by the company went Sequenom. This test, which can only be ordered through a physician, involves blood being taken from the expectant mother, as early as 10 weeks of gestation, and relies on the detection of cell free DNA that circulates between the fetus and the expectant mother. According to the latest research, this blood test can detect up to 98.6% of fetuses with trisomy 21. A 'positive' result on the test means that there is a 98.6% chance that the fetus as trisomy 21; a 'negative' result on the test means that there is a 99.8% chance that the fetus does not have trisomy 21. The turnaround time for the test is about 8-10 days and approximately 0.8% of patients did not receive a result due to technical standards." (13) "Knowing about a birth defect before birth may help you get ready emotionally to care for your baby." (5) "Some women feel that having more information about the baby's health will help them prepare better for the baby's arrival." (18) "It [cell-free fetal DNA testing] is the most accurate screening test for aneuploidy available today." (17) "First trimester screening leads to the detection of approximately 83% Down syndrome cases and 80% of trisomy 18 cases." (25) "The harmony tests has been shown to have detection rate of up to 99% and false positive rate as low as 0.1% for trisomy 21, 18 and 13." (29)

Criteria	Explanation
	• Are potential harms mentioned? "Diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) are accurate for detecting fetal trisomy, but they are invasive and pose a slight risk for fetal loss." (29) "Are there any risks to having an amnio? Some women find that having an amnio is painless. Others feel cramping when the needle enters uterus or pressure when the fluid is removed. One to 2 out of 100 (1 to 2 percent) have cramping, spotting when we gain amniotic fluid after the test. Your provider may tell you to avoid intense activity and take it easy for the rest of the day. Serious complications from amniocentesis are rare. However, the test does involve a small risk of miscarriage. According to ACOG, less than one in 200 women (less than 1 percent) have a miscarriage after amniocentesis." (5) "The doctor will take steps to make the risks of the CVS as low as possible, but no procedure is completely risk-free. The major risk from CVS is miscarriage. A miscarriage may occur if an infection start uterus, if the placenta separates from the uterus, or if the sac surrounding the baby doesn't heal after the procedure, allowing amniotic fluid to leak out. Without CVS, about 2% to 3% of pregnancies will miscarry between 8 and 14 weeks. CVS adds an extra 1% (1 out of 100) risk of miscarriage. Said another way, out of every 100 CVS procedures done, 99 women (99%) do **not** have a miscarriage. Early studies of CVS raised the concern that it increases the risk of birth defects of the fingers and toes. This risk is not increased if a CVS is done after 10 weeks of pregnancy." (18) • Is it mentioned that information gained may impact future pregnancy management decisions? "Some parents want to have a prenatal diagnosis so that they can discontinue their pregnancy. Parents should discuss this option with their obstetrician." (10) "Parents have different reasons for wanting to know about their child's health. Some plan to terminate the pregnancy if the fetus is seriously unhealthy. Others want to prepare for the challenge. For certain medical conditions, the fetus can be treated during pregnancy." (12) "After receiving a prenatal diagnosis in learning more about Down syndrome, some families opt to terminate." (13)

Criteria	Explanation
	"Having a diagnosis allows parents to make choices, such as choosing an adoption plan or stopping the pregnancy." (18)
Potential financial implications and conflicts of interest disclosed	• Are financial costs discussed? "Does insurance cover this test? The list price for the test is $795. With the additional screening for the X and Y chromosome, the price is $815. Unfortunately, we have no information about whether insurance companies will cover the cost or the amount of coverage. We strongly recommend that the patient contact her insurance provider directly to determine coverage, including deductibles and co-pays." (22)

Findings and Interpretation

Ease of Access

Despite the fact that all of the electronic resources are publicly available online, these materials are sometimes as physically difficult to access as printed ones, in that they require knowledge of website or a hyperlink. Some are not directly searchable. Some are in clinician controlled environments, like embedded within electronic medical record systems, pamphlets available only to clinicians who are members of particular professional societies or within health systems intranet servers. Because most popular search engines utilize browser history and demographics to determine what results user receives, the placement of a particular result among other results varies; different people may some results may have a higher placement within the results list than others. In fact, some people may have poor quality results or results from unreliable sources above higher quality or more reliable results. Given that most information seekers likely want the best available information, this seems potentially problematic.

Resources distributed by national obstetrics/gynecology and public health organizations, as well as national government organizations, often require searching within the organization's website, in order to locate the material. When seeking health information, most people use a search engine, such as Google or Bing; these search engines use typically use algorithms to sort pages based upon popularity and relevance. While such an algorithm may correlate well with satisfaction, it is unclear whether it also correlates with high quality health information. For example, the general public may use the term Down syndrome, while professional organizations may use trisomy 21 or chromosomal abnormality; this difference would lower the relevance of the page. Pages not viewed often rank lower than those that are, lowering the popularity. Taken together, health professional organizations have to use lay language, in order to improve their pages relevance, and they need to increase the viewership by sharing a direct link to resources, in

order to increase their popularity. Otherwise, popular websites that already do this, like WebMD will receive more views, regardless of the quality. We might imagine that web browser could alter their algorithms for health related searches, so that popularity and relevance are interpreted in ways that yield information from more reputable sources. However, as this analysis shows, assuming that health education materials distributed by health organizations may not improve the quality of information.

In order for the public to access such resources, they would have to know of the organization's existence and that such materials might be distributed from them. If access to reputable sources is difficult, then we might suspect that prospective parents seeking information about such tests may end up accessing information from questionably reputable sources. It may be valuable for health professionals to identify valuable or good sources and direct patients to them. By increasing the popularity of reputable sites, valuable information will become easier to identify using popular web browsers. As increasing amounts of information is available to prospective parents seeking, health professionals should stay current and up-to-date about the information that this group may be accessing, so that they can ensure patients are adequately and accurately informed.

Based upon the materials assessed, some clinics provide such education materials in web-based formats. While this might facilitate access, patients still must obtain a hyperlink to the resource from their healthcare provider. In efforts to make access to such resources easier for patients and providers, we may need to consider more holistic prenatal health education efforts. For example, rather than different providers offering different web-based resources for specific issues, we might consider creating a pregnancy module, from a reputable source, that gives

access to an array of important health education materials. Then, patients could go to one electronic place to search for topics of interest or obtain trimester-specific guidance.

Information about Related Concepts

One of the most notable barriers to patients accessing up-to-date information is the fact that many of these resources, particularly clinic-based ones, do not specify the date of creation or the date of last update. This relatively simple addition may help ensure patients are basing their decisions off the most recent evidence and serve as a reminder to clinics about when relevant, frequently used health education materials may warrant revisions or updates when the evidence changes.

Maternal age is most commonly described as a risk factor for Down syndrome. Personal history in cases of translocation is also mentioned by some, but to a much lesser extent. As the examples illustrate (Table 5), most materials use very similar or stock language to communicate this risk information. Because of this, the source of the information has little impact on the materials' quality. For example, health education materials from a seemingly reputable health professional organization or nonprofit does not actually seem to generate original content, but instead, they use similar stock language as other seemingly less reputable sources. As a result, the reputable status may not translate into more accurate materials. While accurate, this risk factor information seems more useful before pregnancy, given that such risks are only modifiable before pregnancy (i.e., age and personal history cannot be changed). Because most of the health education materials analyzed here seem to target those contemplating prenatal testing during pregnancy, in contrast to those contemplating pregnancy, these facts about risk factors may be problematic and stigmatizing. Because age itself is not as reliable a predictor of Down syndrome as currently available testing methods, patients should definitely consider a screening option or a combination of options, if patients are concerned about carrying a fetus with a chromosomal

anomaly. The information from a screening test is better to assess the risk of carrying a fetus with a chromosomal anomaly than the patient's age alone. Based upon this information, some might infer that delaying childbearing is the cause of Down syndrome. At a population-level, there is an association between advanced maternal age and Down syndrome; yet, nondisjunction events can occur at any age. We might imagine that more tailored health education materials would mitigate stigmatizing older parents, particularly women, and acknowledge that delaying childbearing happens for a variety of reasons, often beyond individual control. If the intent of discussing maternal age as a risk factor for Down syndrome is to inform parents about Down syndrome risk in subsequent pregnancy, then it should be more clearly stated, and arguably in different health education material.

When discussed, genetics education is conveyed similarly across the various sources of health education materials. Stock language or at least highly replicated language communicates basic concepts about Down syndrome, particularly with regards to descriptions about nondisjunction events and translocations. Because of this, clinic-based health education materials are similarly accessible and adequately describe genetics concepts as those from advocacy organizations. Interestingly, OB/GYN sources and public health ones poorly explain genetics concepts, overlooking or omitting any explanations. Instead, they tend to focus on maternal age as a risk factor for Down syndrome, which may stigmatize older women who are having children[10].

[10] As public comments to NPR radiocasts highlight (See Chapter 4), the public has potential misunderstanding and misconception about age-related risk. Comments illustrate mother-blaming, suggesting that women choosing to delay pregnancy for career goals is the cause of increasing Down syndrome prevalence. I think that this problematically shifts attention to individual decisions, overlooking the lack of societal support for women to have children and pursue career goals simultaneously. In addition, this emphasis on age-related risk for Down syndrome does not acknowledge the potential benefits of being an older parent, like the knowledge and resources that may improve other health outcomes for themselves and their children.

Besides discussion of risk factors, which may be interpreted in a potentially stigmatizing way, much of the health education materials portray Down syndrome in similarly problematic ways. Frequent use of language suggestive of an individual problem −"birth defect", "disorder", and "mental retardation"− are common across most sources. While these word choices reflect disciplinary and/or medical language, they tend to paint Down syndrome in a negative light, because these word choices have largely negative connotations in the public vernacular. For some, this information may be informative, providing information to prospective parents about some common biophysiological outcomes for people with Down syndrome; however, it also focuses almost entirely upon biomedical outcomes, largely overlooking other outcomes, including relationships with family members, friends, and involvement in other social activities. While the information about biophysiological outcomes may be predominately accurate, few of the health education materials direct readers to further information about the claims or provide citations connecting these claims to scholarly literature. Because almost none of the health materials provided when they were last updated, there are limited mechanisms to identify whether the information is up-to-date. Furthermore, while some of the materials mentioned that Down syndrome manifests in diverse ways and to varying degrees, none of the materials provide any more specification than that. For those who would make different pregnancy management decisions based upon particular outcomes, the potential severity and likelihood of associated health conditions might also be valuable; however, this information is not mentioned in most of the health education materials analyzed. As such, we might hope that better health education materials would help to create realistic expectations, discussing potential and likely outcomes for people with Down syndrome and their families. Based upon this analysis and its methods, I cannot determine whether health education materials omit these potentially relevant facts or

whether sufficient evidence simply does not exist based upon current research. If this is entirely the latter, then it seems that in order for prospective parents to be adequately informed about Down syndrome, further investigation about health conditions and quality of life for people with Down syndrome is warranted. Alternatively, we might imagine that some health education developers do not think that such information is valuable or relevant to decisions about prenatal testing. To the best of my knowledge, current research does not indicate whether this is consistent with prospective parents' perceived needs and wants about information during prenatal testing decisionmaking, suggesting the need for more research about potential user needs and wants from information related to these processes.

Information about Testing Methods

The majority of materials focus on a particular testing methodology (e.g., integrated screening, chorionic villus sampling or amniocentesis), minimizing the need for discussion about different testing methods. Problematically, this means the patient must understand the potential decision-making pathways and providers should offer information about such methods in a particular sequence, so that patients are fully informed about the larger decision-making process (i.e., a particular result may lead to further screening or testing or a pregnancy management choice). I do not think that this needs to be a complex matrix or flowchart, but rather I think that patients need to be able to clearly distinguish whether a particular test that they are considering is screening or diagnostic testing.

Most sources discuss testing's purpose, but do so minimally. For example, several note that the purpose of testing is to identify chromosomal abnormalities, like Down syndrome. Within this selection of health education materials, few explicitly discuss why such testing is offered and/or why some women might accept or reject this offer. In other words, why might it be beneficial or not to identify Down syndrome prenatally? For some, it might be valuable to

have quotes from other women or professionals commenting about why they chose the pathway they did. Because this has the potential to be interpreted as leading or directive, I would understand why some health education developers might be reluctant to include this information. Alternatively, I think health education materials might provide greater transparency about these prenatal tests by making comparisons to other existing, optional services; for example, I think prenatal testing for Down syndrome might be compared to prenatal sex identification, where some choose to get this information and others do not.

Unfortunately, when screening and diagnostic methods are differentiated, sources typically only explain that diagnostic methods offer more definitive information than screening methods. I think that more needs to be said. Specifically, I think prospective test-takers need to know that with definitive information from a diagnostic testing some people choose pregnancy continuation, adoption placement, or pregnancy termination. Patients should also know that for some this decision is difficult, so choosing to delay diagnosis until after birth is an option to consider.

Many materials note that testing is voluntary or a personal choice. Clinic-based health education is more likely to mention this in comparison to other sources. This may be because some of the clinic-based materials appear to act as informed consent documents, rather than simply health education. As such, most materials that mention choice refer to choices about screening and/or diagnostic testing. Several mention that some choose to stop a pregnancy following a prenatal diagnosis; however, it is unclear from the descriptions how a choice about a screening test might inform a subsequent choice about diagnostic testing or pregnancy management. Some of these materials paint Down syndrome as negative or unhealthy, suggesting that disability is justification for termination. For some, this information may be

informative, because it illuminates that decision-making about screening and/or diagnostic testing may ultimately inform decisions to terminate a pregnancy; however, for others, such a framing oversimplifies an often difficult, complex decision-making process. Better health education materials might at least acknowledge that few make decisions to end a pregnancy lightly and that decisions based upon testing result should be well-informed. At the very least, I think health education materials ought to be straightforward, specifying that before accepting testing patients considering prenatal screening should identify the risks, benefits, and impact on decisions if the test resulted in a "positive" screening indicative of a higher likelihood of a Down syndrome-affected pregnancy or a "negative" screening indicative of a lower likelihood of a Down syndrome-affected pregnancy.

Options and Potential Outcomes

Testing accuracy is the most commonly mentioned benefit, but a few materials also suggest information as a personal benefit. Given that, at present, all diagnostic methods are invasive, most sources discuss the potential harms to the fetus associated with particular techniques (e.g., pregnancy loss associated with chorionic villus sampling and amniocentesis).

Financial Implications and Conflicts of Interest

Authorship is largely unclear for almost all health education materials. However, it is evident that at least some of the health education materials are produced by the testing companies themselves. It appears as though testing companies offer this information to providers along with testing kits or in conjunction with a testing service. Given the selection bias inherent to purposive sampling and the small number of included studies, a sub-analysis of these materials is not conducted here; however, it may be an important direction for future research. It seems somewhat problematic that testing companies who profit from testing utilization should be tasked with creating health education materials, as an inherent conflict of

interest seems to exist. To the general public, the health education material may not appear to have a commercial interest and it is not explicitly disclosed, except potentially as the name of the test.

Despite much public attention to cost-effective healthcare services, health education almost entirely overlooks financial considerations. Only one source mentions testing costs, and the cost mentioned is for the testing itself. Interestingly, a testing company produced this material. Health education should provide patients with more information about the financial costs of such services, especially for emerging methods that may not be covered by insurance. In addition, health education materials should delineate the costs associated with recommended pre- and post-test counseling, given that they should be provided with such testing but may not be covered by insurance companies that do not cover all forms of prenatal testing.

Patient Supports

Clinics and advocacy organizations provide information that is accurate, well-described for a lay audience, and detailed. For some receiving this information, the volume may seem overwhelming; while for others, the material may be informative and desired. In other words, most of the health education material has been created in a one-size-fits-all fashion with seemingly little consideration for the diversity among people receiving such information. Given that these materials may be offered to a range of learners with highly variable levels of baseline knowledge about prenatal testing for Down syndrome, one of the critical considerations needed for those creating such material is how to tailor and customize the information in such a way that it is maximally useful to the individual. We might imagine that adaptive learning modules in web-based or kiosk-based formats may be an important direction for such materials moving forward. I envision that such adaptive modules would guide patients through scenarios, in order for them to recognize their potential reactions and intuitions. While current research explores

decisional conflict using scenarios (Muller & Cameron, 2015), I propose that patients would have the opportunity to go through similar scenarios, in order for them to better understand their values and the implications of a value-consistent choice without having actually made their own healthcare decisions. This might aid patients in considering alternatives and testing implications without using valuable time within the clinical encounter. Such a tool might also collect information related to misinformation or knowledge needs; for example, additional questions about risk assessment or genetics might help identify patients who will need more time or resources to fully understand testing results and its implications. In this way, patient knowledge, values, and desires from the information will be best addressed through health education materials.

Several materials fail to direct patients to their physician or health professional for further information or clarification. Despite growing amounts of health information available on the Internet, surprisingly few materials direct readers to resources from health professions organizations, advocacy organizations, or government organizations. As this analysis illustrates, this may be because several of the materials from professional and government organizations are of lesser accessibility and quality than other publicly available resources. This is an important area for future work, especially as health education materials are developed for a wider array of genetic conditions and related tests. Rather than creating new materials every time for different conditions or tests, we might envision some resources dedicated to vetting and organizing health education materials for a wide array of conditions warehoused on a single website or web portal, where patients and providers can access accurate, up-to-date health education materials. I foresee that such a resource would more likely be created as part of the efforts of a nonprofit or advocacy organization, as opposed to a national government organization; for example, we might

imagine Genetic Alliance, March of Dimes or Planned Parenthood addressing this topic as it relates to the existing goals of these organizations. In this manner, the organization could target the information related to its mission and goals; for example, Genetic Alliance might focus on discussion the genetics and its inheritance, while Planned Parenthood might focus more on legal access to termination services in particular states at what gestational week in pregnancy. To me, the availability of diverse patient resources is also beneficial, in that patients will have access to health information that offers differing opinions, perspectives, and support. I think that this diversity helps patients access a wider range of information and knowledge upon which to base their decisions, and at least for some, I think this information will empower patients to make choices that are more consistent with their values than most current health materials offer. This would reduce redundant efforts and improve the information available to patients and providers. At the very least, we might imagine similar quality health education materials be available to providers and embedded within electronic medical record systems, where health education materials could be included as part of patient discharge orders.

LIMITATIONS

The major limitation of this project relates to external validity. While the health education materials in this analysis come from a variety of sources, the collection is based upon convenience sampling. While materials were solicited from all time zones, the majority of included health materials are created or provided from the West Coast institutions. This selection bias may have imported particular ideologies and values about prenatal testing. I make no claims that the included sample is representative of other sources, of particular disciplines or advocacy organizations as a broad category, or of all health education materials available on the topic. However, the work does show some of the variability with regards to accessibility and quality that exists in currently available health information. These insights may help health

professionals create informative, accurate, patient-centered health education materials on the topic.

CONCLUSION

In general, there is little difference between clinic-based versus web-based health education materials. The content quality has less to do with where the material is offered and more to do with the sources themselves. Based upon this analysis, the National Down Syndrome Society, an advocacy organization, offers the best health education materials based upon the evaluation criteria used in this analysis. Perhaps interestingly for health professionals, the communication about risk and explanation of genetics are superior to many clinic-based and public health ones −based on the criteria assessed in this analysis− including government produced and distributed ones. The National Down Syndrome Society education materials are more readily accessible than other sources, despite its origination from an advocacy organization. Broader issues related to potentially problematic or stigmatizing language about particular reproductive choices and disability tend to be similar regardless of the source.

In order to improve health education materials about prenatal genetic testing, interprofessional collaboration between genetic counseling, public health, and primary care providers is needed. Inclusion of community-based and advocacy organizations in the development process of such materials, as well as in the dissemination, may help to give prospective parents the needed time and resources to better support decision-making. Broader health education efforts addressing genetics may help facilitate informed consent and empower prospective parents to make the best decisions for themselves and their families.

As the delivery of health education materials evolves and consumers increasingly rely upon the Internet for health information in healthcare decisions, health professionals may need to

consider alternative mechanisms for education and dissemination. While genetic counselors may target particular information and their messages to a particular patient or a specific set of circumstances (e.g., prenatal counseling sessions versus postnatal counseling sessions), public health professionals may not have this ability. While arguably much effort is behind the growing amounts of health education information available to prospective parents about prenatal testing for Down syndrome, it may be valuable to identify already existing, good sources and direct patients to, rather than creating new material. Furthermore, more attention should be directed to user design, in order to ensure that patients have wanted information in reasonable amounts and at appropriate times. In particular, more time and attention addressing preconception counseling seems important, especially when considering risk factors that are unmodifiable during pregnancy, but potentially modifiable before pregnancy. Given the relatively few women who receive preconception counseling, as well as the ease and privacy associated with online health education materials, the creation of evidence-based online resources related to reproductive health may be valuable. We also might imagine health systems or health professionals creating or sponsoring content development and dissemination. Alternatively, we might imagine patient or health advocacy organizations collaborating with health professionals to generate wanted health information in these formats, as a supplement to currently existing health materials.

References

Bakke, A. (2013). Disabling the Rhetoric of Prenatal Genetic Testing: A Critical Analysis of Online Self-Help Literature. *Critical Disability Discourse, 5*, 11-38.

Fox, S. (2012). Pew Internet: Health. Retrieved 24 January, 2013, from http://www.pewinternet.org/Commentary/2011/November/Pew-Internet-Health.aspx

Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. *Qual Health Res, 15*(9), 1277-1288. doi: 10.1177/1049732305276687

Kellogg, G., Slattery, L., Hudgins, L., & Ormond, K. (2014). Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing. *Journal of genetic counseling, 23*(5), 805-813. doi: 10.1007/s10897-014-9694-7

Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing. *Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70*(1), 7-9.

Lawson, K. L., Carlson, K., & Shynkaruk, J. M. (2012). The Portrayal of Down Syndrome in Prenatal Screening Information Pamphlets. *J Obstet Gynaecol Can, 34*(8), 760-768.

Lewis, C., Silcock, C., & Chitty, L. S. (2013). Non-invasive prenatal testing for Down's syndrome: pregnant women's views and likely uptake. *Public Health Genomics, 16*(5), 223-232. doi: 10.1159/000353523

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. *Sociology of health & illness, 32*(1), 37-56.

Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf

Moorhead, S. A., Hazlett, D. E., Harrison, L., Carroll, J. K., Irwin, A., & Hoving, C. (2013). A New Dimension of Health Care: Systematic Review of the Uses, Benefits, and Limitations of Social Media for Health Communication. *Journal of Medical Internet Research, 15*(4), e85. doi: 10.2196/jmir.1933

Muller, C., & Cameron, L. D. (2015). It's complicated – Factors predicting decisional conflict in prenatal diagnostic testing. *Health Expectations*, n/a-n/a. doi: 10.1111/hex.12363

Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. *Health Expectations, 11*(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x

Press, N., & Browner, C. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.

Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.

Reed, K. (2009). 'It's them faulty genes again': women, men and the gendered nature of genetic responsibility in prenatal blood screening. *Sociology of health & illness, 31*(3), 343-359. doi: 10.1111/j.1467-9566.2008.01134.x

Stapleton, H., Kirkham, M., & Thomas, G. (2002). Qualitative study of evidence based leaflets in maternity care. *BMJ : British Medical Journal, 324*(7338), 639-639.

van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. *Patient Educ Couns, 63*(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007

> Newspapers originate and gather much of the newsworthy information that other platforms, like radio and YouTube, distribute. As such, I expect that newspapers influence how these other platforms articulate messages about prenatal testing and diagnosis. While we saw that clinical practice guidelines silence talk about abortion, I suspect that radio import their frames directly from newspapers ─ framing pregnancy termination as a mechanism to prevent disability.
>
> In this section, I illuminate how newspaper articles frame prenatal diagnosis of Down syndrome; I pay particular attention to the modes of testing and screening that the articles reference. In addition, I assess the extent to which these articles adequately address existing concerns and portray the application accurately.

Chapter 4 NEWSPAPER

Amniocentesis or chorionic villus sampling (CVS) to diagnosis Down syndrome has long been an exemplar for prenatal genetic testing (President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, 1983). In her foundational disability rights critique of this practice, Asch argues that unreflective use of prenatal testing may erode reproductive autonomy by providing information that can transform a wanted pregnancy and child into an unwanted one (Asch, 1999, 2000; Asch & Wasserman, 2005). This and subsequent Disability Studies literature focus on how negative attitudes, assumptions, and misunderstandings about living with a disability and its implications for family may lead prospective parents to make incompletely informed choices about selective termination (Hubbard, 2010; Kittay & Kittay, 2000; Klein, 2011; Saxton, 2010; Taylor, 2013; Tremain, 2006). In contrast, much of the bioethics literature is relatively silent about prospective parents understanding of disability, and instead, they focus on whether pregnant women are sufficiently informed about the purpose of testing, understanding the risks and benefits, and potential testing outcomes within the clinical encounter (Etchegary *et al.*, 2008; Lippman & Wilfond, 1992; Potter *et al.*, 2008; Press & Browner, 1994; Press & Browner, 1997a, 1997c; St-Jacques *et al.*, 2008; van den Berg, Timmermans, Ten Kate, van Vugt, & van der Wal, 2005, 2006; van den Heuvel *et al.*, 2010). As this scholarship highlights, beyond its intrinsic link to abortion debates,

prenatal diagnosis still raises other issues about autonomy (Davis, 2010; de Jong, Dondorp, Frints, de Die-Smulders, & de Wert, 2011; Lindemann & Nelson, 2008; Markens, Browner, & Preloran, 2010; McCoyd, 2010; Press, 2008; Scully, 2008; Seavilleklein, 2009). While by no means an exhaustive list, these continuing conversations about prenatal testing highlight the diversity and nuance of viewpoints regarding its evolving practice.

Informed consent is undoubtedly a central issue with regards to prenatal testing, but looking only at how patients are informed within the clinical encounter seems insufficient to broad understanding about their choices. Here, I extend that analysis to information they may receive from newspapers. Because it represents a culmination of advancements in understanding about fetal DNA circulating in maternal blood and DNA amplification technologies, Lo's "Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis" (1998) marks a substantial turning point in the public conversation. With this proof-of-concept publication, the clinical application of noninvasive prenatal diagnosis using cell-free fetal DNA testing emerged. Committee opinions from esteemed health professions ushered in a new era of prenatal genetic tests utilizing this methodology. These tests may someday diagnose a far broader range of genetic conditions, but at present, they are often used like other screening tools (i.e., maternal serum screening and ultrasound) with follow-up diagnostic test (i.e., amniocentesis or chorionic villus sampling) (Devers *et al.*, 2013; Langlois *et al.*, 2013; The Noninvasive Prenatal Screening Work Group, 2013). These scientific developments and clinical advisories have drawn much media attention. As a dominant source of information about medical developments and emerging technologies, media plays an important role in informing the public (Dolan, Iredale, Williams, & Ameen, 2004). Given that the average American accesses news from multiple sources, including television, newspapers,

and social media (Media Insight Project *et al.*, 2014), media representations will likely shape public knowledge about this emerging technology.

Yet, the existing literature also highlights many concerns about media representations. With regards to news about biomedical developments, some question how completely and adequately news about new treatments report the benefits, risks, and costs (Moynihan *et al.*, 2000; Schwitzer, 2008; Wilson, Bonevski, Jones, & Henry, 2009). In attempts to quantify risks and benefits, journalists sometimes poorly explain the meaning behind the numbers in a way that is understandable for their audience (Gigerenzer, Gaissmaier, Kurz-Milcke, Schwartz, & Woloshin, 2007). Particularly with regards to genetic research, most news accurately conveys the results and scientific claims articulated in scientific journals, but it places greater emphasis on benefits, minimizing risks (Bubela & Caulfield, 2004). Some of this "genohype" −overly optimistic framing of genetics research− may result from inaccurate representations by journalists and/or overstatements by scientists (Caulfield, 2004; Holtzman *et al.*, 2005; Shuchman & Wilkes, 1997). At the same time, relatively little media attention is given to the quality of life of people living with these very conditions that such testing commonly targets. Yet, Disability Studies critiques have long highlighted that people with disabilities are framed as having an inherent problem needing medical intervention to cure or fix, objects of pity or inspiration, and infantilized (Haller, 2010; Mills & Erzikova, 2012; Riley, 2005; Shapiro, 1993), which in this context may misinform prospective parents about living with disability and its implications for family. If these media frames emerge in news about cell-free fetal DNA testing for Down syndrome, then we expect that the public may have misinformation when entering the clinical encounter.

Given the volume of information received from media sources, we bring together some of the existing ethical concerns about informed consent along with media critiques about representations of emerging technology and disability. We acknowledge that the current media landscape is evolving: shifting towards digital platforms, changing consumption habits, and varying levels of public trust and skepticism in these sources (Media Insight Project et al., 2014; Pew Research Center, 2010a, 2012). However, during the timeframe of interest to this analysis, newspapers create much of the content that other platforms, like radio and blogs, distributed (Pew Research Center, 2010c). Therefore, I illuminate how newspaper articles inform their audience, including potential patients who might encounter cell-free fetal DNA testing and healthcare providers who offer it. I assess the extent to which these articles adequately address existing concerns, including accurately portraying the practice of prenatal testing and the experience of living with Down syndrome. Because of how these representations may influence a broad audience, we discuss how a sample of news articles creates what we consider to be unrealistic expectations about this methodology in use as a clinical application.

METHODS

NEWSPAPER SELECTION

We searched LexisNexis Academic for newspaper articles. Given that many of the contextual issues related to prenatal testing, like clinical practice guidelines, healthcare policies and practices are country-specific, we restricted our analysis to English language, US-based newspapers where the full-text newspaper articles were available (N=219). We excluded newspapers that did not have such articles available during the timeframe of interest (n=49) and topic specific newspapers (n=42, e.g., Advertising Age, California Energy and Climate report, Home and Textiles). A purposive sample of 10 newspapers (The New York Post, USA Today, The Spokesman-Review (Spokane, Washington), The Philadelphia Inquirer, Los Angeles Times,

The New York Times, The Denver Post, The Washington Post, The Baltimore Sun, St. Louis Post-Dispatch) was selected based upon top circulation (Lulofs, 2013) and geographic region, in order to ensure that all U.S. time zones were included.

Article Selection

Given the timeframe in which cell-free fetal DNA testing was developed, we restricted our search to articles published in English between 1997 and 2013. We included articles regardless of type (e.g., editorials, news, business, etc.), length, and placement. The specific terms used in the search were: (((prenatal W/3 screen) OR (prenatal W/3 test) OR (prenatal W/3 diagnosis) OR amniocentesis) and Date (geq(12/31/1997) and leq(09/01/2013))). This broad approach identified 447 articles. This broad search strategy ensured that potentially relevant articles were not excluded. The full-text of each article was downloaded.

In order to find the most relevant articles about prenatal testing for Down syndrome in the United States, a set of specific inclusion criteria were applied. We deemed articles that did not fit these criteria as beyond the scope of this study, and such articles were excluded from further analysis.

1. The primary or secondary purpose of the article addressed cell-free fetal DNA testing.
2. Down syndrome (trisomy 21) had to be discussed or identified in the text. Because Lo's (1998) work and practice guidelines (Devers et al., 2013; Langlois et al., 2013; The Noninvasive Prenatal Screening Work Group, 2013) specifically address Down syndrome, we restricted our search to this. We recognized that such technology has potentially broader applicability (Kitzman *et al.*, 2012). However, during the earliest part of the timeframe in which potentially relevant articles were published, there was limited evidence about whether such testing would be realistic and/or appropriate for other genetic conditions in clinical practice (Devaney, Palomaki, Scott, & Bianchi, 2011; Palomaki *et al.*, 2011).
3. Articles were only included once. Duplicates or articles re-published in a different edition were excluded, so that no single article weighted the results in a particular direction.

By assessing the title and full-text of each article, two independent coders reviewed each article. When discrepancies arose, both independent coders returned to the original full-text article together and discussed the article; through conversation about the original data, they reconciled these discrepancies and came to consensus about our determination to include or exclude an article. Based upon this review, 424 articles (223 did not address the testing method of interest; 52 did not address Down syndrome; 149 were duplicates or reprints) were excluded for not meeting these criteria. Most articles were excluded because they did not address the testing method of interest; they often reported sonography developments, discussing this technology or integrated panels (using both ultrasound and maternal serum testing). In total, 23 articles were included for analysis (Table 6).

Data Analysis

Using a checklist developed specifically for this study (Table 7), two independent coders rated each article. The checklist criteria are based upon literature in bioethics and health journalism and tailored to address issues that may arise in reporting of noninvasive prenatal testing. Each article was assessed on each item. Items were marked as "strongly disagree", "disagree", "neutral", "agree", "strongly agree", or "not applicable". The initial inter-rater reliability was assessed using a weighted Kappa statistic (κ=0.80), indicating a good level of agreement. When discrepancies arose, both independent coders returned to the original full-text together and discussed the article passages that led them to their classification; through conversation about relevant passages, they reconciled these discrepancies and came to consensus about their categorical determination. Using the Proofing tool in Microsoft Word™, the Flesch–Kincaid Grade Level and the Flesch Reading Ease were calculated to assess reading level.

Using a set of guiding questions (Table 8), the analysis employed a form of directed content analysis (Hsieh & Shannon, 2005). We qualitatively explored concepts addressed within

the checklist, in order to illustrate judgments about how the newspaper articles discuss: the purposes of testing, the benefits, harms, and costs; the methodology, explaining who should use the testing; as well as key actors and their potential conflicts of interest. For this study, we did not have an *a priori* list of assumptions about purposes of testing, the benefits, harms, and costs of this emerging technology; instead, we relied upon the articles to illustrate what newspapers framed as purpose, benefits, harms and costs. The unit of analysis was each individual newspaper article. The newspaper, publication date, section, title, author, and full-text article were imported into ATLAS.ti 7™ for analysis.

FINDINGS

A detailed accounting of the article assessment is provided in Table 9, and a sample of illustrative quotes supporting these judgments about how the newspapers portrayed the criteria is provided in Table 10. We looked for the article to articulate the purpose(s) of testing. The majority of articles (70%) accurately describe (A, agree and strongly agree) the purpose of prenatal genetic screening as a means to detect and/or to diagnose specific genetic conditions. Some of the other purposes the newspaper articles articulate are: trait identification (e.g., paternity, genetic conditions, sex), preparation, risk assessment, and pregnancy management.

Table 6. Included newspaper articles (in chronological order by publication date)

Identification Number	Article
1	The Associated Press. 2000. Down syndrome test could become safer. The Denver Post, A-16.
2	News Services and Staff Reports. 2004. FINDINGS. The Washington Post, A12.
3	Pollack, Andrew. 2008. Blood Tests Ease Search For Down Syndrome. The New York Times, D5.
4	Ganguli, Ishani. 2008. Blood Test Might Identify Down Syndrome. The Washington Post, HE02.
5	Stein, Rob. 2009. New Safety, New Concerns In Tests for Down Syndrome. The Washington Post, A01.
6	Huget, Jennifer LaRue, and anti-elitist. 2009. The Checkup; Health in the News and in Your Life (Adapted from: voices.washpost.come/checkup). The Washington Post, HE02.
7	Hurley, Dan. 2011. "All I Could Think Is, She's My Baby, She's A Lovely Girl And What Can I Do To Help Her?". The New York Times, Section MM28.
8	Roan, Shari, and Los Angeles Times. 2011. Prenatal Blood Test Detects Down Syndrome. *Spokesman Review* A; Pg. 3.
9	Pollack, Andrew. 2011. A Less Risky Down Test Lifts Hopes. The New York Times, Section B1.
10	Skotko, Brian. 2011. Will America cull people with Down syndrome?; A new, simpler test just 10 weeks into a pregnancy can identify the genetic disorder. Then, a difficult choice. USA Today, 9A.
11	Bernhard, Blythe. 2011. Prenatal test raises concerns Accurate, safer screening for Down syndrome may prompt abortions, some say. St. Louis Post-Dispatch, A1.
12	Krieger, Lisa M., and San Jose Mercury News. 2012. Identifying birth defects early creates new questions; Blood test finds genetic details of fetus weeks into pregnancy. Spokesman Review, A1.
13	Pollack, Andrew. 2012. Tests Of Parents Are Used To Map Genes Of A Fetus. The New York Times, Section A1.
14	Doughton, Sandi, and Seattle Times. 2012. UW researchers map genes of unborn child. Spokesman Review, A4.
15	Douthat, Ross. 2012. Eugenics, Past and Future. The New York Times, Section Sunday Review Desk, 12.
16	Pollack, Andrew. 2012. Before Birth, Dad's ID. The New York Times, Section B1.
17	Pollack, Andrew. 2012. Clinical Trial Is Favorable For a Prenatal Gene Test. The New York Times, B1.
18	McCullough, Marie. 2012. Seeing a fetus' future ills; The power, and perplexity, of prenatal testing. The Philadelphia Inqu*irer*, A01.
19	Rubin, Rita. 2012. New fetal test creates dilemma for some women. The Washington Post, E01.
20	Lloyd, Janice. 2013. Advances in medicine are on Halle Berry's side; Being 46 and diabetic add to pregnancy risks. USA Today, 6D.
21	Lewis, Lloyd, and Julie Reiskin. 2013. The rise of a new eugenics. Denver Post, 26A.

Identification Number	Article
22	Szabo, Liz. 2013. Earlier prenatal tests usher in "heartbreaking" decisions; Advocates for those with Down syndrome tell the world these lives are worth living. USA Today, 1A.
23	Winerman, Lea. 2013. New prenatal tests reveal far more about fetuses. The Washington Post, E06.

Table 7. Assessment criteria

	Criteria	Qualitative questions	Rationale
	An article that realistically and accurately portrays noninvasive prenatal diagnosis using cell-free fetal DNA testing:		
A	Articulates the purpose(s) of such testing	What purposes were described? How so?	The article should clearly demonstrate that this approach does not ensure their future child's health (Press & Browner, 1994; St-Jacques et al., 2008).
B	Adequately addresses potential benefits	What benefits were described? How so?	The article should convey the magnitude of potential benefits (Schwitzer, 2008; Wilson, Bonevski, Jones, & Henry, 2009).
C	Adequately explains potential harms	What harms were described? How so?	The article should portray an accurate understanding of the potential harms of the approach (Schwitzer, 2008; Wilson, Bonevski, Jones, & Henry, 2009).
D	Adequately discusses financial costs	What financial costs were described? How so?	The article should highlight the costs of the new approach, including the extent to which insurance would cover such approaches (Schwitzer, 2008; Wilson, Bonevski, Jones, & Henry, 2009).
E	Compares application with existing methods	What comparisons were discussed? How so?	The article should directly compare this new approach to amniocentesis or chorionic villus sampling (Schwitzer, 2008; Wilson, Bonevski, Jones, & Henry, 2009).
F	Explains all statistics	What statistics were provided? How were they explained?	The article should make numbers easily understandable to the reader (Gigerenzer, Gaissmaier, Kurz-Milcke, Schwartz, & Woloshin, 2007).
G	Reports the target population	How was the target population described?	The articles should highlight that health professions committees emphasize that cell-free fetal DNA testing is not appropriate for all pregnant women, only those who are at high risk for particular conditions (Committee Opinion No. 545, 2012).
H	Seeks out a variety of viewpoints	Who is represented as a key actor?	Journalists create "experts" to demonstrate balanced coverage, but this polarizes issues that have far more complexity than two-sides (Hivon, Lehoux, Denis, & Rock, 2009), so articles should include a variety of viewpoints.
I	Discloses potential conflicts of interest	How are potential conflicts disclosed?	Commercial pressures and lack of independent sources may lead to hype of new approaches (Agarwal, Sayres, Cho, Cook-Deegan, & Chandrasekharan, 2013; Moynihan, Heath, & Henry, 2002).
J	Is comprehensible for a general audience (reading level)		

Each item was marked as "strongly disagree", "disagree", "neither agree nor disagree", "agree", "strongly agree" or "not applicable"

Table 8. Newspaper analysis guide

Guiding Questions
Stakeholders • What key actors are involved in the discourse? • What disciplinary discourses enter the conversation? • Who is represented in the discourse?
Situated meaning • How do word-specific choices create particular meanings?
Cell-free fetal DNA testing • How is testing discussed? • What benefits/concerns are expressed? Who expresses them? • How is this testing distinguished from invasive diagnostic tests? • How is this testing distinguished from noninvasive screening tests?
Clinical utility • How is clinical utility of noninvasive prenatal testing expressed? • What is the purpose(s) of such testing? • What counts as clinical utility, when not explicitly discussed?
Down syndrome • What models of disability dominate the discourse? • How is the medical or social model of disability expressed? • Do other models of disability enter the conversation?
Eugenics • How is eugenics discussed? • How do different actors talk about it? • What actors silence eugenics talk?
Choice • How is choice used? • What choices are being articulated or discussed? • How do different actors talk about it? • What actors silence choice talk?

Table 9. Article evaluation by criteria (in percentage)

Criteria	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
A	9	22	0	52	17
B	0	22	4	22	52
C	26	4	13	35	22
D	39	0	30	17	13
E	4	4	35	44	13
F	62	24	14	0	0
G	56	13	0	17	13
H	30	4	17	0	48
I	13	9	17	39	22
J	The average Flesch–Kincaid Grade Level is 12.5 (range 10.5 – 15.9) and the Flesch Reading Ease is 42.7 (range 26.7-54.8).				

Table 10. Samples of quotes

Criteria	
A	"to diagnose Down syndrome, which causes mental retardation and physical abnormalities" (1) "to terminate if the fetus lacked a favorable trait" (13) "detect a 'disability' or 'genetic disorder'" (21) "give doctors and parents an early window on the fetus and enable medical teams to anticipate complications" (22)
B	"new tests are designed to offer more definitive results early in the pregnancy." (5) "near 99% accuracy" (10) "to further reduce the number of women who undergo amniocentesis" (11) "the power to reveal a wide range of potential problems before birth" (14)
C	"'The capacity of genomics to generate data is outstripping our ability to interpret it in useful ways'." (Jay Shendure quoted in 14) "Is this sort of 'liberal eugenics'' in which the agents of reproductive selection are parents rather than the state, entirely different from the eugenics of Fisher's era, which forced sterilization on unwilling men and women?" (15) "it is not always possible to tell whether a small abnormality detected […] will be harmful to a child, or if so, how severe such a problem will be." (17) "This information - Bernhardt called it "toxic knowledge" - led to anxiety, confusion, frustration, depression, and, in some cases, abortion. (Barbara A. Bernhardt referred to in 18)
D	"a list price of $2,000 but that the real cost insurers would pay would be $600 to $800. That is more than existing screening tests but less than amniocentesis." (3) "The test is expected to cost about $1,900, about as much as amniocentesis [...] privately insured women would have to pay $235 out of pocket" (9) "researcher estimate that [the test] would cost $20,000 to $50,000 to do one fetal genome today." (13) "New medical technologies often challenge a health-care industry grappling with pressures to control costs. It's not yet clear whether the new tests will cut costs and miscarriages by reducing invasive prenatal diagnostic procedures such as amniocentesis or inflate cost" (19)
E	"more accurate than the blood and ultrasound screenings currently used to gauge the chance that a baby will be born with the syndrome" (11) "while most existing prenatal tests are designed to check for single disorders, including Down syndrome, a full-gene scan has the power to reveal a wide range of potential problems before birth" (14)

	"a new blood test is a safer method to detect birth defects than amniocentesis (drawing amniotic fluid from the uterus), which carries a slight risk of miscarriage." (20)
F	"Doing a procedure on 1 percent is way better than doing it on 5 percent or 10 percent of all people" (3) "scientists would have expected a 34 percent increase in the number of babies born with the syndrome between 1989 and 2005. Instead, 15 percent fewer were born." (6) "That study on pregnancies at high risk for Down syndrome found a detection rate of 98.6 percent (209 of 212 pregnancies)." (8) "In 90 percent of cases, a positive test for Down syndrome leads to an abortion." (15)
G	"For pregnant women worried about Down syndrome and other chromosomal abnormalities in their unborn child, a new blood test could ease their minds" (4) "women were selected for the research trial if they were considered high-risk due to advanced age, if their fetus tested positive through conventional tests for chromosomal defects, or if they previously had a baby with chromosomal defects." (12) "the American College of Obstetricians and Gynecologists recommends the test only for women who are 35 or older or have other risk factors such as a family history of one of the disorders, because not enough studies have examined its effectiveness in low-risk women." (23)
H	"'The geneticists expect Down syndrome to disappear,' Costa says, 'so why fund treatments?'" (Alberto Costa quoted in 7) "it's a search-and-destroy mission, where the baby is aborted, we are not in favor of it." (12) "Ravgen, a small company in Columbia, MD, has been offering its test on a limited basis and charges $950 to $1,650, depending on the circumstances, said Dr. Ravinder Dhallan, the chief executive." (16) "There is an urgent need for policymakers, regulators and professional societies to provide guidance on the most efficient and ethical manner for such tests to be introduced." (18)
I	"The test was developed by Ravgen Inc., a private biotechnology company based in Maryland that reported the results in this week's Journal of the American Medical Association." (2) "Dr. Brian G. Skotko of the Down syndrome program at Children's Hospital Boston. His sister has Down syndrome" (9) "Lloyd Lewis is board chair of the Rocky Mountain Down Syndrome Association. He is CEO of Arch Thrift stores and president of the Colorado Cross Disability Coalition." (21)

The articles raise an array of benefits and harms related to such tests, including both those directly related to the test and broader implications. We looked for the article to adequately address potential benefits and harms, where higher rated articles provided at least two examples for each. Nearly three-quarters of the articles (74%) discuss the benefits of this cell-free DNA testing (B, agree and strongly agree), including accuracy (thirteen articles), availability during first trimester (eleven articles), more information (nine articles), safety (eleven articles), and timely results (two articles). A substantial portion of articles (57%) also explain the potential harms of testing (C, agree and strongly agree), including too much or confusing information (twelve articles), eugenics (nine articles), accuracy (seven articles), emotional distress (six articles), testing leads to abortion (five articles), industry or regulatory involvement (five articles), and timing (three articles). We assessed whether articles compared this emerging technology to existing methods. If the article suggested that cell-free fetal DNA should be used for diagnostic purposes, it needed to compare this application to amniocentesis and/or CVS, the "gold" standard for clinical diagnosis; alternatively, if the article suggested that cell-free fetal DNA testing should be used for screening purposes, it needed it compare this application to ultrasound or maternal serum screening. We considered general comparisons to be acceptable, including invasiveness of testing, accuracy, or timing of testing offer. Broadly, the benefits and harms are often (43%: E, neutral, disagree and strongly disagree) described without direct comparisons.

We assessed if articles discussed the financial implications of testing, either through insurance or private costs to the consumer. Articles that simply provided a price without justification rated lower than those that gave both price and explanation. Cost and financial responsibility is framed as a benefit (e.g., in 9) and a concern (e.g., in 19). Sometimes, the cost

is just stated (30%: D, neutral) without any reasoning or not reported (39%: D, disagree and strongly disagree). Thirteen articles (57%) discuss the commercial underpinnings of such testing, drawing upon larger conversations about cost-effective healthcare services, even though only four articles (17%) are located in the Finance/Business sections.

We looked for articles to describe an appropriate population who might receive testing; at the time the newspaper articles were published, cell-free fetal DNA testing was only deemed clinically appropriate for at-risk groups, including, for example, those with a personal or family history of Down syndrome. Those that distinguished at-risk groups rated higher than articles that did not specify a target population. Relatively few articles (30%: G, agree and strongly agree) discuss that cell-free fetal DNA testing is most appropriate for women with "at-risk" pregnancies, due to personal or family histories of Down syndrome births or due to age. We explored whether articles provide any numerical or quantitative evidence throughout the text. This included false-positive or false-negative rates, Down syndrome birth prevalence, or pregnancy termination rates. Articles that provided no explanation rated lower than those that explained all numerical evidence. Twenty articles (86%) fail to describe or explain their statistics (F, disagree and strongly disagree): they left numerical values without indicating the meaning (e.g., in 3); used different denominators to express risk (e.g., in 20); used confusing language that might muddle any interpretation of the statistics (e.g., in 8); and referenced findings from scientific literature without explanation of relevant details (e.g., in 22).

We reviewed the articles for inclusion of diverse viewpoints as represented by quotes or references to external sources. Articles rated higher if they provided more than one perspective and higher still if they included more than a scientific or medical perspective. Most articles include an array of viewpoints. Slightly more than half of the articles (12) included two or fewer

perspectives (H, neutral, disagree and strongly disagree); however, the main perspectives represented were from scientists and medical professionals. Twenty articles (86%) include quotes or discussion from scientific researchers, medical professionals, or related professional organizations (e.g., Albert Costa, Jacob Kitzman, Joe Leigh Simpson, the American College of Medical Genetics, and the American Congress of Obstetricians and Gynecologists). We looked across all articles for all the represented agents and compared their disclosed conflicts of interest. Three articles (13%: I, strongly disagree) omit relevant financial relationships, identified in other articles. Seven articles (30%) represent expert knowledge by including insights from scholars with academic appointments or affiliations, but who did not have biomedical training or expertise. The articles frame these individuals as having expertise because of their background or experience addressing ethical, legal, social implications of genetic testing (e.g., Ruth Faden, Hank Greely, Peter Singer, Erik Parens, and Marcy Darnovsky). To a lesser extent, community members were included as experts. Six articles (26%) represent people with Down syndrome and/or their families, but this category often arises when actors identified with dual expertise. For example, the articles highlight Brian Skotko, a physician and sibling of a person with Down syndrome (e.g., in 5, 20, or 22), and Michael Bérubé, a humanities scholar and parent of a child with Down syndrome (e.g., in 7). Nine newspaper articles framed women as the sole reproductive decision makers (in 5, 6, 7, 10, 12, 16, 17, 19, 22), particularly with regards to pregnancy termination choices. These women were not specifically discussed as experts of their bodies, choice, or proxy authorities for their developing fetuses. While the label of reproductive decision-maker endows women with the right to choose and the responsibilities associated with these choices, this framing problematically suggest that such choices are primarily individualistic, overlooking familial or relational considerations often embedded in these

decisions. The audience is another key stakeholder with regards to newspaper representations, albeit largely silent within the newsprint conversation (only 1 article included comments from readers). The average Flesch–Kincaid Grade Level (J) is 12.5 (range 10.5 – 15.9) and the Flesch Reading Ease (J) is 42.7 (range 26.7-54.8), indicating an above high school reading level.

DISCUSSION

The majority of articles accurately portray that the purpose of noninvasive prenatal testing is to identify genetic conditions, including Down syndrome. Only four articles mention that testing results might help people prepare for a child with Down syndrome. Many of these articles discuss Down syndrome in terms of medical issues. Yet, Down syndrome need not primarily be considered in this way; instead, disability might framed as human diversity (Amundson, 2005; Amundson & Tresky, 2007) or as identity (Garland-Thomson, 2005; Linton, 1998). This is simply to say that there are multiple ways to frame disability and that broader inclusion of more diverse representations may help provide the public a more complete understanding of Down syndrome. The manner in which these newspaper articles frame disability may ultimately skew readers' understanding about the purpose of noninvasive prenatal tests. Newspapers skillfully articulate multiple purposes of testing, but some readers may interpret these purposes as reasons one should test, rather than as reasons one could test. We support this information about multiple purposes in so far as it helps prospective parents consider why they might accept such testing. However, in conjunction with largely negative framing of Down syndrome, we suspect that at least some people may benefit from having health education materials or informed consent documents that explicitly state: rejecting testing, delaying testing, or waiting until birth for more information are valid options, as well. Media representations overlook what for many is a difficult process and decision between prenatal testing, prenatal

diagnosis, and pregnancy termination. Ensuring that patients know the range of available options and giving them greater control over the timing of these options may promote informed consent within the clinical encounter.

Newspapers articulate both the benefits and harms of noninvasive prenatal testing, discussing an array of potentials for each. Often times, however, benefits were described at greater length and frequency than the harms (Table 9, B and C). To the reader, this imbalance may suggest that the benefits outweigh the harms. This representation of noninvasive prenatal testing parallels health journalists' critiques that reporting tends to overemphasize benefits, overlooking harms (Schwitzer, 2008; Wilson et al., 2009). We might expect that such framing could influence individuals' assessment and decision-making about utilization of this testing, in that people may be more willing to accept tests that they perceive as beneficial.

Comparisons to existing prenatal screening and testing practices also serve to highlight potential benefits and harms of noninvasive testing. Most commonly, this comparison is made to amniocentesis and chorionic villus sampling. However, some newspapers also make comparisons between maternal serum screening and/or ultrasound. For the most part, these comparisons are helpful, and readers can better understand the difference between emerging noninvasive prenatal tests and current standards of care. Most reader confusion likely arises when newspaper articles inadequately name the test or series that they are comparing to, especially given the notable differences between prenatal screening (i.e., maternal serum screening and/or ultrasound) and prenatal diagnostic testing (i.e., amniocentesis or chorionic villus sampling).

While the majority of the articles mention cost, a large portion (39%) do not report or explain it. Even of those that do, few do so well. As previous assessments suggest (Schwitzer,

2008), information about costs and financial responsibility are inadequate: information about cost without mention of insurance; information about cost with certain insurance; broad estimates; no comparison of costs to existing screening and testing methods. This incompleteness seems largely to result from the complexity of healthcare costs and spending, but at the same time, readers cannot reasonably make cost-value assessments of such testing.

The poorly explained statistics is the most prevalent issue across these articles. Some of the common issues relate to statistics about testing accuracy, Down syndrome birth rates, and pregnancy termination rates following prenatal diagnosis. These issues with statistics seem related to incompleteness, rather than explicit inaccuracy. Most of the articles appropriately report that noninvasive prenatal testing is accurate. They offer limited description about how such tests are more accurate than existing screening methods, failing to explain the jargon used (e.g., cases, false-positive or false-negative) or translate the original study for a lay audience. Perhaps, in an attempt to provide clarity, some articles repeat percentages as numbers (Table 10, F). Regarding birth rates, the articles provide insufficient contextual descriptions, in order to adequately inform the reader (e.g., in 6). The articles do not specify whether a crude or adjusted (for age and/or ethnicity) birth rate is referenced, or even what geographic region or study population is included. These details might shape readers' understanding about the statistic and ultimately even their perspective about the article's claims (e.g., when the statistic is used to suggest whether noninvasive prenatal testing is eugenic or not). Several articles (n=5) made reference to pregnancy termination following prenatal diagnosis, offering a 90% statistic. While accurate, none make reference to any empirical evidence grounding this proposed fact - despite discussion within scholarly literature (Mansfield, Hopfer, & Marteau, 1999; Natoli, Ackerman, McDermott, & Edwards, 2012). Furthermore, none explain contextual issues related to this

statistic: this includes that termination rates vary by condition and by geographic region; findings are based upon research studies with a relatively small sample size, rather than population-level data; findings are based upon invasive procedures with known risk of spontaneous pregnancy loss, which may exclude some people who choose to continue pregnancies but opt-out of this testing.

The contextual incompleteness of the statistics may result from the demand for short length (Holtzman et al., 2005), but in doing so, the ability of an article to be informative may be compromised. As Gigerenzer, Gaissmaier, Kurz-Milcke, Schwartz & Woloshin (2007) highlight, nontransparent framing of statistics −usually unintentionally− results from a lack of understanding on the part of the author, but may also −usually intentionally− result from attempts to persuade or manipulate. While this nontransparent framing of statistics may result from author misunderstanding, we also want acknowledge that it may result from author bias. Noninvasive prenatal testing raise a host of hotly contested issues, including abortion, eugenics, disability, medical and social costs, etc. We expect that health news reporting be as balanced as possible; however, journalists have particular viewpoints, like everyone else, which may enter their reporting unintentionally via their use and framing of statistics.

The majority of articles did not specify for whom the testing is most appropriate. While American College of Obstetricians and Gynecologists recommends offering screening for conditions like Down syndrome to all pregnant women (ACOG Office of Communications, 2007), they issued distinct guidance about cell-free fetal DNA testing, stating that at the time, it was only appropriate for at-risk groups (Langlois et al., 2013). By poorly acknowledging this important limitation of the evidence at the time of their publication, the articles imply that cell-free fetal DNA testing is appropriate for all pregnant women. Similar to disease mongering

−what health journalism critics describe as medicalizing human variations in health (like social phobia, osteoporosis, and erectile dysfunction) to expand the market of a particular product (Moynihan, Heath, & Henry, 2002)− this simple omission makes testing seem relevant to a wider audience, in terms of interest and clinical applicability. Consistent with work addressing a connection between prenatal testing and medicalization of pregnancy (Inhorn, 2006; Ivry, 2009; Lyerly *et al.*, 2007; Rapp, 1999), we interpret this framing of noninvasive prenatal testing as reflective of a trend towards medicalization, given the absence of disease or even risk for many pregnant women.

Nearly half of the articles include a variety of viewpoints. The relatively limited engagement of people with Down syndrome and their families as experts -without medical, scientific, or otherwise scholarly expertise- gives authority about Down syndrome to scientists and clinicians. With the exception of Dr. Brian Skotko, who is a sibling to someone with Down syndrome, all of the family members included are parents. Parents are key actors, given that the experience of prenatal testing is theirs; however, the fact that none of the articles include parents of adults with Down syndrome infantilizes people with Down syndrome. Prospective parents receiving a prenatal diagnosis may be concerned with their early parenting experiences, and newspapers do highlight this time point. However, the articles' focus on children with Down syndrome may constrain readers' imagination about lifelong outcomes for people with Down syndrome. People with Down syndrome are not completely absent from other forms of media and some readers may have personal knowledge and experience that people with Down syndrome can live happy, productive lives; however, the dominant media framing tends to focus the public's attention to childhood, perpetuating a frame of dependency (Baglieri & Shapiro, 2012). The general lack of readily available, circulating information about living with Down

syndrome is problematic, because the public may have limited or misinformation about quality of life and appropriate expectations for people with Down syndrome across the lifespan. Their arguably limited knowledge may influence testing behavior and pregnancy decisions, so emphasis on lifelong outcomes for people with Down syndrome within the clinical encounter may be justified. A more complete representation of viewpoints to include people with Down syndrome valued as experts for their lived experience across the lifespan. A full representation of viewpoints should include also include those who choose to terminate their pregnancies following a prenatal diagnosis, so that people who may want to make similar choices can draw upon their experiential knowledge and first-hand experience, too.

External validity is a major limitation of this project. The regional contexts may be different from other parts of the country and other countries, especially with regards to abortion, science education, and mass media. We make no claims that this sample of newspaper articles is representative of other US newspapers, of the individual newspapers, or of the authors' or audiences' views. Furthermore, the database and search terms used may have created selection bias. There was not a specific word count that needed to discuss noninvasive prenatal testing to be included; the analysis gave equal weight to articles that briefly mentioned such testing and those where the entire discussion was devoted to this topic in its entirety. As such, some articles may be discounted or rated lower for not providing a variety of perspectives about prenatal testing, when in fact, prenatal testing was not fully explored. This analysis only included the text, ignoring potentially important cues that images may suggest. We did not give more weight to articles based upon number of readers or print date (e.g. New York Times articles did not receive more weight in our analysis than ones from St. Louis Post Dispatch, or articles published on Sunday did not receive more weight in the analysis than those published on Monday). Our

assessment of conflicts of interest is based upon conflicts identified in other articles; therefore, it is possible that we under-represented the number of conflicts due to omissions made across all articles. Nonetheless, the work does show how newspapers reflect truths about the purpose of such testing, as well as an array of potential benefits and harms; the work also highlights some potential issues that arise with the framing of noninvasive prenatal testing.

CONCLUSION

The analysis suggests that newspaper articles about noninvasive prenatal testing have some room for improvement in how they represent this emerging technology and clinical application to the public. As is, the health messages received from reading newspapers may confuse prospective parents, who are already faced with tough reproductive choices, about the value of noninvasive prenatal testing. While the majority of articles articulate the purpose and even purposes of testing, there is much emphasis on abortion and eugenics. By making a direct connection between testing and pregnancy termination, newspaper articles overlook a decision making process –screening, noninvasive prenatal testing, diagnostic testing and potentially pregnancy termination or continuation– that might be better illuminated by including people who chose to terminate pregnancies following prenatal diagnosis as experts to talk about their experiences.

For the most part, newspapers do a great job of explaining the benefits of noninvasive testing, but harms are not explained, as well. Because of this imbalanced representation, readers may have insufficient evidence upon which to assess whether such testing is beneficial or harmful. Furthermore, our analysis suggests that many of these benefits and harms are crafted without directly comparing noninvasive prenatal testing to existing standards of care. Rather than simply telling the readers what is good and bad about noninvasive testing, we think it would

be better to illustrate the differences between it and existing methods, so that readers can come to their own conclusions about what is a benefit and harm.

The cost and financial responsibility of healthcare is confusing, but increasingly important in an environment with pressures to control spending. As such, articles should consider reporting the cost of the standard of care, so that the reader can make comparisons, as well as relevant factors that might alter the price or financial responsibility reported and to what end (e.g., falling sequencing prices might reduce prices by a specific amount or insurance coverage might reduce out-of-pocket costs by a specific amount).

In addition, articles should clearly explain for whom such testing is most appropriate. By suggesting that noninvasive testing is best for a wider audience than the scientific or clinical guidelines do, newspaper representations can appear to be expanding the market of the application. Similarly, statistics are by and large poorly explained – lacking important details that might influence how the reader interprets the data. This incompleteness in these articles may cause the readers to be unknowingly "sold" particular ideas about noninvasive testing, as opposed to educated with balanced coverage.

While articles do a good job including a variety of viewpoints, much of the attention focuses on scientists and clinicians. This is perhaps concerning given that several of the experts have relevant financial relationships that were not disclosed; namely, articles omitted patent holdings and ownership in biotechnology companies profiting from genetic testing platforms for some of the included medical and scientific experts. Including more coverage of people with Down syndrome and their families –across the lifespan− may help to provide readers more holistic information.

Our findings highlight that newspapers are circulating information that is inconsistent with clinical guidance. These findings should raise the bar with regards to our understanding about informed consent, in that patients may need to unlearn and re-learn information before making important healthcare decisions. Because the reading level of these articles is so high, we might suspect that newspapers framing will have the greatest impact on college-educated readers, including clinical providers. Ultimately, this analysis may help guide readers to critically evaluate the issues being presented in newspapers. Similarly, it may alert clinicians involved with the offer of such testing to potential misunderstandings that patients may have based on newspaper representations. By ensuring that patients understand the complexities of such testing, health professionals can engage and empower patients to make informed choices best suited to their individual and familial needs.

As noninvasive prenatal testing is translated into clinical practice more readily, it may be beneficial for clinical providers and future research to assess pre-visit knowledge about noninvasive testing and to determine how such knowledge was accessed. In this manner, clinicians can help patients unlearn misinformation that may influence decision-making and ensure that patients have access to accurate, up-to-date information, in order to best formulate their decisions. While the focus of this project was on noninvasive testing for Down syndrome, we recognize the potential of such testing to be used for a wider array of genetic conditions; in this manner, more viewpoints may need to enter the public conversation. Further research and media attention needs to explore the experiences of people who face this clinical application −including people who choose to test and those who do not, as well as those who choose to terminate and those who do not− in order to highlight the range of experiences and variety of options. We hope this creates newsworthy stories that can provide more balanced information to

those making these reproductive choices and can educate providers about how to guide patients during this often difficult time.

References

ACOG Office of Communications. (2007). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

Amundson, R. (2005). Disability, Ideology, and Quality of Life: A Bias in Biomedical Ethics. In D. T. Wasserman, R. S. Wachbroit & J. E. Bickenbach (Eds.), *Quality of life and human difference : genetic testing, health care, and disability* (pp. 101-124). New York: Cambridge University Press.

Amundson, R., & Tresky, S. (2007). On a Bioethical Challenge to Disability Rights. *Journal of Medicine and Philosophy, 32*(6), 541-561. doi: 10.1080/03605310701680924

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.

Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. *Prenatal testing and disability rights, 51*.

Asch, A., & Wasserman, D. (2005). Where is the sin in synecdoche. *Quality of life and human difference: Genetic testing, health care, and disability*.

Baglieri, S., & Shapiro, A. (2012). *Disability studies and the inclusive classroom: Critical practices for creating least restrictive attitudes*: Routledge.

Bubela, T. M., & Caulfield, T. A. (2004). Do the print media "hype" genetic research? A comparison of newspaper stories and peer-reviewed research papers. *Canadian Medical Association Journal, 170*(9), 1399-1407. doi: 10.1503/cmaj.1030762

Caulfield, T. (2004). Biotechnology and the popular press: hype and the selling of science. *TRENDS in biotechnology, 22*(7), 337-339.

Davis, D. S. (2010). Genetic Dilemmas: Reproductive Technology, Parental Choices, and Children's Futures.

de Jong, A., Dondorp, W. J., Frints, S. G., de Die-Smulders, C. E., & de Wert, G. M. (2011). Advances in prenatal screening: the ethical dimension. *Nature Reviews Genetics, 12*(9), 657-663.

Devaney, S. A., Palomaki, G. E., Scott, J. A., & Bianchi, D. (2011). Noninvasive fetal sex determination using cell-free fetal DNA: a systematic review and meta-analysis. *JAMA, 306*(6), 627-636.

Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2013). Noninvasive prenatal testing/noninvasive prenatal diagnosis: the position of the National Society of Genetic Counselors. *Journal of genetic counseling, 22*(3), 291-295.

Dolan, G., Iredale, R., Williams, R., & Ameen, J. (2004). Consumer use of the internet for health information: a survey of primary care patients. *International Journal of Consumer Studies, 28*(2), 147-153.

Etchegary, H., Potter, B., Howley, H., Cappelli, M., Coyle, D., Graham, I., . . . Wilson, B. (2008). The influence of experiential knowledge on prenatal screening and testing decisions. *Genetic testing, 12*(1), 115-124.

Garland-Thomson, R. (2005). Disability and Representation. *PMLA, 120*(2), 522-527.

Gigerenzer, G., Gaissmaier, W., Kurz-Milcke, E., Schwartz, L. M., & Woloshin, S. (2007). Helping doctors and patients make sense of health statistics. *Psychological science in the public interest, 8*(2), 53-96.

Haller, B. A. (2010). *Representing disability in an ableist world : essays on mass media*. Louisville, KY: The Advocado Press.

Holtzman, N. A., Bernhardt, B. A., Mountcastle-Shah, E., Rodgers, J. E., Tambor, E., & Geller, G. (2005). The quality of media reports on discoveries related to human genetic diseases. *Public Health Genomics, 8*(3), 133-144.

Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. *Qual Health Res, 15*(9), 1277-1288. doi: 10.1177/1049732305276687

Hubbard, R. (2010). Abortion and disability: who should and who should not inhabit the world. In L. J. Davis (Ed.), *The Disability Studies Reader* (3 ed., pp. 107-119). New York, NY: Routledge.

Inhorn, M. C. (2006). Defining women's health: A dozen messages from more than 150 ethnographies. *Medical Anthropology Quarterly, 20*(3), 345-378.

Ivry, T. (2009). *Embodying culture: pregnancy in Japan and Israel*: Rutgers University Press.

Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: Conversations with my son. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights* (pp. 165-195): Georgetown UP.

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. *Science translational medicine, 4*(137), 137ra176-137ra176.

Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". *AJOB Primary Research, 2*(2), 8-20.

Langlois, S., Brock, J.-A., Wilson, R. D., Audibert, F., Carroll, J., Cartier, L., . . . Senikas, V. (2013). Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. *J Obstet Gynaecol Can, 35*(2), 177-181.

Lindemann, H., & Nelson, J. L. (2008). The romance of the family. *Hastings Center Report, 38*(4), 19-21.

Linton, S. (1998). *Claiming disability : knowledge and identity*. New York: New York University Press.

Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. *American journal of human genetics, 51*(4), 936.

Lo, Y., Tein, M. S., Lau, T. K., Haines, C. J., Leung, T. N., Poon, P. M., . . . Hjelm, N. M. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. *The American Journal of Human Genetics, 62*(4), 768-775.

Lulofs, N. (2013). Top 25 U.S. Newspapers for March 2013. Retrieved 08-27, 2014, from http://www.auditedmedia.com/news/blog/top-25-us-newspapers-for-march-2013.aspx

Lyerly, A. D., Mitchell, L. M., Armstrong, E. M., Harris, L. H., Kukla, R., Kuppermann, M., & Little, M. O. (2007). Risks, values, and decision making surrounding pregnancy. *Obstetrics & Gynecology, 109*(4), 979-984.

Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. *Prenatal diagnosis, 19*(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. *Sociology of health & illness, 32*(1), 37-56.

McCoyd, J. L. (2010). Authoritative knowledge, the technological imperative and women's responses to prenatal diagnostic technologies. *Culture, Medicine, and Psychiatry, 34*(4), 590-614.

Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf

Mills, C. B., & Erzikova, E. (2012). Prenatal Testing, Disability, and Termination: An Examination of Newspaper Framing. *Disability Studies Quarterly, 32*(3).

Moynihan, R., Bero, L., Ross-Degnan, D., Henry, D., Lee, K., Watkins, J., . . . Soumerai, S. B. (2000). Coverage by the news media of the benefits and risks of medications. *New England Journal of Medicine, 342*(22), 1645-1650.

Moynihan, R., Heath, I., & Henry, D. (2002). Selling sickness: the pharmaceutical industry and disease mongeringCommentary: Medicalisation of risk factors. *Bmj, 324*(7342), 886-891.

Natoli, J. L., Ackerman, D. L., McDermott, S., & Edwards, J. G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). *Prenatal diagnosis, 32*(2), 142-153.

Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine, 13*(11), 913-920.

Pew Research Center. (2010a). Americans Spending More Time Following the News. Retrieved 08-26, 2014, from http://www.people-press.org/files/legacy-pdf/652.pdf

Pew Research Center. (2010c). How News Happens. Retrieved 08-26, 2014, from http://www.journalism.org/2010/01/11/how-news-happens/

Pew Research Center. (2012). Where Americans Get News. Retrieved 08-26, 2014, from http://www.journalism.org/media-indicators/where-americans-get-news/

Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Boland, I. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. *Health Expectations, 11*(4), 355-365.

President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. (1983). *Screening and Counseling for Genetic Conditions: A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs*. Washington, DC: U. S. Government Printing Office Retrieved from http://bioethics.georgetown.edu/pcbe/reports/past_commissions/geneticscreening.pdf.

Press, N. A. (2008). Genetic Testing and Screening. *From Birth to Death and Bench to Clinic: The Hastings Center Bioethics Briefing Book for Journalists, Policymakers, and Campaigns*, 73-78.

Press, N. A., & Browner, C. H. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.

Press, N. A., & Browner, C. H. (1997a). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.

Press, N. A., & Browner, C. H. (1997c). Why women say yes to prenatal diagnosis. *Social Science & Medicine, 45*(7), 979-989.

Rapp, R. (1999). *Testing women, testing the fetus: The social impact of amniocentesis in America*.
Riley, C. A. (2005). *Disability and the media: prescriptions for change*. Hanover, NH: University Press of New England.
Saxton, M. (2010). Disability rights and selective abortion. In L. J. Davis (Ed.), *The Disability Studies Reader* (3 ed., pp. 120-132). New York, NY: Routledge.
Schwitzer, G. (2008). How do US journalists cover treatments, tests, products, and procedures? An evaluation of 500 stories. *PLoS medicine, 5*(5), e95.
Scully, J. L. (2008). Disability and genetics in the era of genomic medicine. *Nature Reviews Genetics, 9*(10), 797-802.
Seavilleklein, V. (2009). Challenging the rhetoric of choice in prenatal screening. *Bioethics, 23*(1), 68-77.
Shapiro, J. P. (1993). *No pity : people with disabilities forging a new civil rights movement*. New York: Times Books.
Shuchman, M., & Wilkes, M. S. (1997). Medical scientists and health news reporting: a case of miscommunication. *Ann Intern Med, 126*(12), 976-982.
St-Jacques, S., Grenier, S., Charland, M., Forest, J. C., Rousseau, F., & Légaré, F. (2008). Decisional needs assessment regarding Down syndrome prenatal testing: a systematic review of the perceptions of women, their partners and health professionals. *Prenatal diagnosis, 28*(13), 1183-1203.
Taylor, A. (2013). " Lives Worth Living:" Theorizing Moral Status and Expressions of Human Life. *Disability Studies Quarterly, 33*(4).
The Noninvasive Prenatal Screening Work Group. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. *Genetics in Medicine, 15*(5), 395-398.
Tremain, S. (2006). Reproductive Freedom, Self-Regulation, and the Government of Impairment in Utero. *Hypatia, 21*(1), 35-53. doi: 10.2307/3811076
van den Berg, M., Timmermans, D. R., Ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2005). Are pregnant women making informed choices about prenatal screening? *Genet Med, 7*(5), 332-338.
van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. *Patient Educ Couns, 63*(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007
van den Heuvel, A., Chitty, L., Dormandy, E., Newson, A., Deans, Z., Attwood, S., . . . Marteau, T. M. (2010). Will the introduction of non-invasive prenatal diagnostic testing erode informed choices? An experimental study of health care professionals. *Patient Educ Couns, 78*(1), 24-28. doi: 10.1016/j.pec.2009.05.014
Wilson, A., Bonevski, B., Jones, A., & Henry, D. (2009). Media reporting of health interventions: signs of improvement, but major problems persist. *PLoS One, 4*(3), e4831.

YouTube is a popular forum for public video-sharing. In this section, I investigate a random sample of YouTube videos with content about prenatal testing for Down syndrome. I examine this collection of YouTube videos as a means to illuminate how the public conveys meaning about Down syndrome and related prenatal testing, as well as who is represented and not in shaping this public discourse. I identify themes and rhetorical patterns across 53 videos, noting some systemic issues with regards to representation, quality content, and corporate involvement.

Chapter 5 YOUTUBE

In the era of Health 2.0[11], people might utilize a variety of websites and applications to create and to share health information online. Of U.S. adults who are Internet users, 72% looked online for health information within the past year; when accounting for those who do not go online, this equates to 59% of the total US adult population (Fox & Duggan, 2013). Such searching is not done in complete isolation. In fact, 39% looked for information related to someone else's health situation, and an additional 15% looked for both information related to their health situation and someone else's (Fox & Duggan, 2013). People connect with other people's experiences online. Nearly a third (26%) of online health information seekers read or watched a video about someone else's health situation or experience in the past year (Fox & Duggan, 2013). Online health information seekers under 50 years of age are more likely to seek information specifically about pregnancy and childbirth; in fact, 12% of adult Internet users sought health information related to this in the past 12 months (Fox & Duggan, 2013). More than half (53%) of online health information seekers say that the information prompts them to

[11] I use the term Health 2.0 as a more encompassing term than Medicine 2.0. In part, I use this language to reflect the fact that these web-based interactions take place explicitly outside of the clinical encounter. While I acknowledge that healthcare professionals and organizations are increasingly engaging patients using these platforms, I consider this more direct interaction to be Medicine 2.0. I also mean to distinguish Health 2.0 (i.e., creating, sharing, communicating, and collaborating about health information via web-based platforms) from Health 3.0 (i.e., tracking, collecting, sharing, and retrieving health information using virtual tools). See Hughes, Joshi, & Wareham (2008) and Lupton (2014) for a more complete discussion.

ask a doctor new questions or to get another healthcare provider's opinion. As such, at least some prospective parents and their family members likely search for information about prenatal testing for Down syndrome online, and specifically, I consider how this shift in information access is transforming community formation, networking, and support. YouTube videos, unlike other sources for health messages, afford the public the opportunity to create a counter-narrative about prenatal testing, disability and selective termination than other media sources. These personal narratives might help better inform those contemplating testing, diagnosis, or selective termination with wanted information that is unavailable or only marginally available from other sources.

Online videos are a major part of the digital landscape. In fact, 72% of all adult US Internet users watch these on video-sharing sites, like YouTube, and these rates are even higher for those under 50 years of age − 92% and 81% for 18 to 29-year-olds and 30 to 49-year-olds, respectively (Fox, 2013). Even if this age group does not seek information about prenatal testing in YouTube videos, we might imagine that YouTube could become a resource for health information, if high quality or otherwise wanted information was available there. The age groups who have high rates of online video watching are the same as those seeking health information about pregnancy and childbirth, and as noted above, health information seekers are searching for information about pregnancy and childbirth online, even if not in YouTube videos. Given this, I explore YouTube videos as a public resource for health communication and education about prenatal testing for Down syndrome. Despite tensions about privacy, many people publically or pseudonymously share health information to gather information, to build relationships with people who have similar life experiences, and to raise awareness about particular health issues.

In recent literature, personal narratives about health shared publicly as YouTube videos have been explored. Chou, Hunt, Folkers, & Augustson (2011) investigate narratives from cancer survivors, noting how most of these narratives focus on the diagnosis; cancer is framed as unexpected and the diagnosis as unforgettable. Cancer survivors create dramatic tension by discussing the mundane and a sense of normalcy of everyday life leading up to the diagnosis. Reported speech or thought, where the narrator imitates voice of themselves or others is a common motif in this storytelling; by doing this, the narrator creates an sense of immediacy about the interaction between the narrator and others, as well as further dramatic tension (Chou et al., 2011). Cancer narratives on YouTube are not limited to adults, as Clerici, Veneroni, Bisogno, Trapuzzano, & Ferrari (2012) point out, and in the pediatric narratives, children and/or their families, as opposed to clinicians, predominantly share these videos − sometimes to commemorate dying and death. The video format and structure seem to make sharing impressions and experiences easier than written text, because the viewer does not have to read and mentally develop the scene, as one would do with reading; instead, the viewer is provided visual cues that place them within the scene. These videos may be used as a resource for other patients coping with similar diseases, because they offer practical and emotional perspectives about information and resources over the course of the disease (Clerici et al., 2012). Yet, ethnic and racial minorities are underrepresented in this venue, which may perpetuate the disproportionate cancer burden that these groups experience because patients do not have access to this potentially useful, culturally-relevant resource and coping tool (Eddens *et al.*, 2009).

As we see in relationship to other conditions, YouTube videos offer a variety of mechanisms of peer support. For people with mental health conditions, including people with schizophrenia, schizoaffective disorder, and bipolar disorder, this video-sharing minimizes

isolation, creates avenues of exchange and reciprocity about coping strategies to everyday issues, as well as medication use and medical intervention (Naslund, Grande, Aschbrenner, & Elwyn, 2014). Many YouTube videos document pre- and post-treatment experiences, demonstrating their symptoms and performing self-administered tests to quantify results; in this manner, patients create so-called experiential-evidence that may be significant and valuable to others experiencing similar conditions (Mazanderani, O'Neill, & Powell, 2013). More specifically to genetics and genetic testing, Harris, Kelly, & Wyatt (2014) call these "autobiologies", referring to the study and story about one's own biology; they illuminate how users of direct-to-consumer genetic tests make sense of their biology by interweaving stories about genetic markers and family histories in playful ways. Despite growing public engagement with health issues in YouTube videos, several authors have expressed concerns about corporate involvement through advertisements and the questionable content quality (Gabarron, Fernandez-Luque, Armayones, & Lau, 2013; Syed-Abdul *et al.*, 2013; Yang, Seo, Patel, & Sansgiry, 2012). While little is known about who and how communities engage with prenatal testing for Down syndrome in public stories about genetics specifically, I consider existing concerns about representation, corporate involvement, and content quality in these videos, as the results elaborate.

By exploring YouTube videos as publically shared video narratives, I identify who is involved and absent from this community. More importantly, I illuminate how community members engage and exchange messages about such testing, highlighting how this platform provides incomplete information, rather than creating a constructive space for personal narratives about prenatal testing decision-making.

METHODS

Using privacy mode to prevent browsing history, networks and friend's recommendations from influencing video retrieval, YouTube was searched for videos; the specific phrase "prenatal testing for Down syndrome" generated 1,750 videos, of which 523 were unique. Non-English language videos and duplicates were excluded. We analyzed a random 10% sample (n=53) (Table 11); each randomized video was analyzed. Each video was recorded using Snagit 11™ and imported into Atlas.ti 7 for analysis. Videos were not excluded on the basis of content, in order to make inferences about the relevance of content available on YouTube about prenatal testing.

Table 11. YouTube videos included for analysis.

Number	Author	Title	Date
1	Melanie Kinsworthy	Is Down Syndrome becoming extinct?	February 11, 2012
2	Jared Buckley	What Causes Down Syndrome?	March 21, 2014
3	IntermountainMoms	What kinds of tests will I have throughout my pregnancy?	April 18, 2013
4	AttitudeLive	The Harper Family – Part Three	June 3, 2008
5	Sanjida Ahmed	Vote of Thanks by Pankaj Sohaney	March 30, 2013
6	IsisParenting	Early Pregnancy Screening: New Research in Non-Invasive Testing	April 9, 2013
7	CdnDownSyndrome	Meet Paul – Canadian Down Syndrome Society	July 4, 2012
8	MPI-CBG	Science Cafe September 2012	October 8, 2012
9	Howdini	How to understand prenatal testing and genetic screening	August 27, 2008
10	CdnDownSyndrome	"See the Ability" with the Canadian Down Syndrome Society (2014)	March 19, 2014
11	BiologyProject1000	Down Syndrome-Genetic Counselling	May 12, 2013
12	Massachusetts Down Syndrome Congress	Norwich woman running Boston Marathon to raise money for Down Syndrome YouTube	April 15, 2014
13	Nikolay's Genetics Lessons	Down Syndrome and Amniocentesis explained	February 15, 2014
14	Hermes Prado Jr	Prenatal test – Amniocentesis – Fetal medicine	February 6, 2009
15	ExpandedBooks	Prenatal Paternity Test – Non-Invasive	February 11, 2014

Number	Author	Title	Date
16	NUScast	2012 The Catalyst – Quick Down's Syndrome test	October 2, 2012
17	CdnDownSyndrome	Lauren Potter is coming to the Canadian Down Syndrome Conference!	April 1, 2013
18	Developmental Disorders	Care For Babies With Down Syndrome Trisomy 21	May 5, 2014
19	Developmental Disorders	How-To Read Down Syndrome Markers On An Ultrasound During Pregnancy	May 5, 2014
20	Brian Skotko	Down syndrome: 99% love their families	January 29, 2012
21	Life Matters TV	Down Syndrome Pregnancies	February 26, 2008
22	Parents Magazine	First Trimester of Pregnancy – Tests and Screening	December 12, 2012
23	Dsgofkc	Down Syndrome Guild of Greater Kansas City	February 18, 2009
24	Anuj13	Prenatal testing pros and cons	April 25, 2010
25	MissingSky101	Fukushima Rad News 3/14/13: 10 million abandoned in high radiation areas after Fukushima	March 14, 2013
26	Heritage Chiropractic YouTube	Abby – Down Syndrome Awareness Week	October 31, 2011
27	Nir Aharoni	Free Sytle improvisation by haim, boy with Down syndrome	November 7, 2008
28	The Center for Bioethics & Human Dignity	Legislation and Policy Regarding an Unexpected Prenatal Diagnosis	October 16, 2013
29	ONExShoTxScott	Pinky the Magic Spinning Ghost Manite Fetus with Down Syndrome of Blackout	March 7, 2009
30	SIforAGE	SIforAGE Science Corner: Mara Dierssen	June 25, 2013
31	Jan87906	Prenatal Testing	December 5, 2013
32	Angieboo504	IVF, Prenatal Screening, Surrogacy	February 25, 2010
33	Midpacific Exploratory	Prenatal Testing Bioethics	May 14, 2013
34	Eric Martin	DSAMC Buddy Walk commercial 2012	September 10, 2012
35	BiggUp	How to prevent birth defects	October 8, 2014
36	Global Down Syndrome Foundation	Hallmark "Home and Family" with John C. McGinley – Part 1	January 17, 2014

Number	Author	Title	Date
37	IFCD PMay	Fragile X Syndrome for Primary Care Physicians	May 10, 2014
38	Ryan Young	Holly's and the Decision to have Brooke	July 28, 2011
39	PHIL100UW	3.5 Prenatal Genetic Screening and Harm to the Child	July 21, 2011
40	Pregnancy Care	Down's Syndrome Test, Amniocentesis test, Ultra Sound Scan in Pregnancy	October 1, 2013
41	The Center for Bioethics & Human Dignity	Managing an Unexpected Prenatal Diagnosis: Conference Introduction	October 16, 2013
42	The Pound Report	A Mini 'THE OFFICE' Reunion at TWENTYWONDER 2013 and MORE! THE BEST OF THE BEST THUS FAR!	July 19, 2013
43	Eric brown	Where To Get Dr J. Mansoor prenatal parenting course Online	October 23, 2012
44	AlexanderFreeTour	What to do when you have "I am enough" abundance	2009
45	Scansurrey	BBC News Article – Combined Test for Down's Screening	May 2, 2009
46	IntermountainMoms	What is non-invasive prenatal testing and who can have that done	January 25, 2013
47	Ten	Down's Syndrome test	September 5, 2012
48	Lake Charles Memorial Health System	Prenatal Genetic Testing with Dr. Gisele McKinney	November 11, 2013
49	Cherishlifevideos	Pregnancy for Downs Syndrome – Wed 13 Feb 2013 (Seven News BNE)	February 14, 2013
50	VCU Life Sciences	On Down – Down Syndrome	June 24, 2008
51	Massachusetts Down Syndrome Congress	Dr. Brian Skotko presenting at the MDSC 28th Annual Conference	March 20, 2012
52	OhioHealth	New ultrasound at Riverside Methodist to Check for Downs Syndrome & other chromosome diseases	July 23, 2009
53	Salomon Says	Testing for Downs Syndrome	April 15, 2002

Using a set of guiding questions (Table 12), the analysis employed a form of directed content analysis (Hsieh & Shannon, 2005). The unit of analysis is the posted video, including narrative, audio and visual representations. The themes are based upon each video's content themes, as well as how the video's content compared to the content themes of other videos.

Based upon the methodology implemented with this analysis, the purpose for uploading a video or creating such information was inferred (i.e., there was no attempt to identify the video generator and contact them to ask about their purpose(s). The analysis qualitatively explored the purposes of testing, the benefits, harms, and costs; disability representations; as well as key actors and their potential conflicts of interest. Through an iterative process, two independent reviewers coded these texts. When discrepancies arose, both independent coders returned to the original full-text together and discussed the article passages that led them to their classification; through conversation about relevant passages, they reconciled these discrepancies and came to consensus about their determination. The codes were built into broad themes.

Table 12. YouTube video analysis guide

Guiding Questions
Stakeholders • What key actors are involved in YouTube videos? • What disciplinary discourses (bioethics, public health and genetic counseling) enter YouTube videos? • Who is represented in YouTube videos? • What issues of representation are noticeable?
Situated meaning • How do word-specific choices create particular meanings?
Cell-free fetal DNA testing • How is testing discussed? Is testing discussed accurately? • What concerns/benefits are expressed? Who expresses them? • How is this testing distinguished from invasive diagnostic tests? • How is this testing distinguished from noninvasive screening tests? • Do YouTube videos reflect evidence-based health practices and policies? • Are biotechnology companies or genetic testing company advertising in YouTube videos? • Are potential conflicts of interest disclosed?
Clinical utility • How is the clinical utility of noninvasive prenatal testing expressed in YouTube videos? • What is the purpose(s) of such testing? • What counts as clinical utility, when not explicitly discussed?
Down syndrome • What models of disability surface in YouTube videos? • How is the medical or social model of disability expressed?

Guiding Questions
Abortion • How is abortion discussed in YouTube videos? • What key actors talk about abortion or silence it? **Eugenics** • How is eugenics discussed in YouTube videos? • How do different actors talk about it? What actors silence eugenics talk? **Choice** • How is choice used in YouTube videos? What choices are being articulated or discussed? • How do different actors talk about it? What actors silence choice talk?

FINDINGS

Each of the 53 videos were grouped into five broad categories (exclusion, educational, community outreach, commercial, and strategic) based upon emerging themes, as opposed to *a priori* classifications. The majority of the available videos (36 videos, 68%) were published between 2012 and 2014. The video generators varied widely, but included disability rights organizations, healthcare organizations or systems, anonymous or pseudonymous members of the public. The content included promotional clips, academic-style conference presentations, recordings of televised or electronic news segments, and homemade video clips or presentations with a narrated compilation of pictures and written factoids.

Six videos (11%) fit in an exclusion category, because they did not address prenatal testing or Down syndrome in a substantive way. If we extrapolate this random sample to the 523 unique videos generated using the search terms discussed, then we would expect that 58 videos would similarly not address prenatal testing or Down syndrome.

Nearly half (n=24, 45%) were categorized as educational. They describe what Down syndrome is and/or symptoms of the condition; alternatively, they discuss prenatal screening and/or diagnostic testing. With this educational category, several mention ethical, legal and social issues associated with prenatal testing and selective termination. Some of the topics they address include eugenics, the value of a child with disability in life generally and specifically for

family life. They mention ethical, legal, and social issues are casually, without fully explaining or discussing. Notably, most of the educational videos were created and/or distributed by people who did not have an academic appointment or scholarly expertise and subject matter. Brian Skokto, obstetrician-gynecologist who has a sibling with Down syndrome, was the only recurring (three times) scholarly expert. A few involved role-playing or imitating patient-provider interactions (e.g., reading a script where someone enacted being patient and someone enacted being a genetic counseling, as in 11). Of these, four targeted healthcare providers, and 20 were directed towards a general public audience; however, the content did not change significantly, regardless of the audience.

Nine videos (17%) were categorized as community outreach. Namely, disability rights organizations seeking funding, advocating for particular causes, or establishing an agenda publicly. The community outreach category is similar to the educational category in terms of images of people with Down syndrome and their families and friends. However, for this analysis, this category is distinct from the educational one in two significant ways: first, all these videos explicitly connect to disability rights organizations, and second, these videos did not specifically address prenatal testing or explicitly related issues, despite being retrieved using the YouTube search query.

Eight videos (15%) were categories as commercials, where a particular organizations advertised information or procedures related to prenatal testing. The organizations included for-profit companies, hospitals and healthcare systems, and pro-life/choice advocacy organizations. The commercials often combined educational efforts with deliberate branding and implicit or explicit referral to services. The content of videos in the commercial category is similar to those in the educational category, but this category is distinct from educational videos in that they were

branded with a particular organization's involvement. The involved organizations have seemingly specific, directive agendas related to prenatal testing or selective termination.

Six videos (11%) were categorized as strategic, attempting to sway viewers' opinion about Down syndrome or prenatal testing and diagnosis. Within this category, commentary from public figures (i.e., using popular celebrities, like Laura Potter who has Down syndrome herself from Glee in 17; John McGinley has a child with Down syndrome from Scrubs in 298); evidence from academicians, clinicians or scholars; or, discussion about morals or religion, is used to elevate particular perspectives. In terms of content, the videos in this category are similar to those in the educational category or community outreach, in that they highlight similar images and narratives of people with Down syndrome and their families and friends; unlike the educational or community outreach oriented videos, these videos are explicit about the purpose and intent of providing such information. Therefore, in terms of style and approach, the videos in this category are similar to those in the commercial category, in that they have a much more directed approach.

DISCUSSION

Parents of children with Down syndrome are by far the most representative group in YouTube videos selected for analysis. Consistent with earlier research about cancer narratives (Eddens et al., 2009), racial and ethnic minorities were largely absent from YouTube videos. While many of the videos categorized as community outreach did include racial and ethnic minorities, these groups were noticeably absent from all the other categories. The absence of racial and ethnic minorities may perpetuate an idea that white families mostly choose to have and to raise a child with Down syndrome or that racial and ethnic minorities mostly choose selective termination. In part, however, this bias may have been introduced by selecting only English

language videos. If there are continued efforts to utilize YouTube videos for educational purposes, then there is need for greater inclusion of racial and ethnic minorities as experts and as participants. Given the diversity of patients who may receive the offer of prenatal testing, greater inclusion may help to ensure YouTube videos reflect a range of the perspectives, attitudes and cultural beliefs. Existing scholarship highlights that race, ethnicity, and education may contribute to and shape decision-making (Farrell, Hawkins, Barragan, Hudgins, & Taylor, 2015; Learman *et al.*, 2003; Rapp, 1999). Therefore, greater diversity in YouTube videos about prenatal testing might improve access to health information reflecting their attitudes, values, and cultural beliefs for those facing these decisions about prenatal testing, selective termination or continuing a pregnancy following prenatal diagnosis.

Children with Down syndrome are commonly represented and even some adults with Down syndrome are seen in the community outreach category. However, people with Down syndrome are largely excluded from the other categories. Particularly with regards to the education category, we might imagine that people with Down syndrome have valuable perspectives and lived experience to share. Unlike any other stakeholder, people with Down syndrome can best inform others about the health conditions they might experience, the everyday joys and challenges, and their general quality of life. This information may be invaluable to those facing decisions about prenatal testing and potentially selective termination; furthermore, we might imagine that posting videos on this topic in such a public space may help to create a nonmedical but accessible resource.

Despite reportedly common termination following prenatal diagnosis of Down syndrome, in all of the YouTube videos in this analysis where a pregnancy management decision was discussed, pregnancies were continued. This means that people who choose termination

following prenatal diagnosis of Down syndrome are underrepresented in YouTube videos. This might be attributable to social desirability bias associated with people not wanting to disclose or share termination experiences publicly, suggesting that pregnancy termination is more taboo and stigmatizing than many other health behaviors and decisions openly shared in these videos. As we might expect from this public video-sharing resource, disciplinary and professional dialogue is largely missing, except when used in commercials.

By and large, the videos in this analysis are incomplete and reflect dated information, despite recent postings. Most of the descriptive information, (i.e., information about what testing particular tests involve and the timing) is portrayed accurately. However, few videos in any category, even the educational ones, articulate what genetic conditions prenatal tests identify. In addition, few videos explicitly discuss the purposes, risks or benefits. As such, clinical utility is not discussed. Taken together, thematically, we might infer from these YouTube videos that clinical utility means personal risks and benefits in the context of our health care system and society; this inference comes from the emphasis on personal choice and repeated discussion of ethical, legal, and social implications of prenatal testing (e.g., eugenics, the value of a child with disability in life generally and specifically for family life, wrongful birth or life tort, etc.). Few videos discuss risk factors that might increase the likelihood of a pregnancy being affected with Down syndrome. Few videos explain the typical serial relationship from screening methods to diagnostic ones or that diagnostic testing is most appropriate for pregnant women at high risk, as identified from screening or personal risk factors. If patients base decisions largely on the content of YouTube videos, then they would not necessarily be misinformed about prenatal testing methods, but rather inadequately informed. Furthermore, it is nearly impossible −even for the author, who has expertise in this area− to distinguish videos produced or shared by people

and/or companies with conflicts of interest. In this manner, typical viewers may not be able to separate educational from commercial videos.

In several of the videos, the medical model of disability is evident, especially in the educational category. However, the vast majority of YouTube videos in this analysis offer a more nuanced framing of disability, suggesting both challenges and value from living with Down syndrome. Down syndrome is portrayed as part of life, which is suggestive of a diversity model of disability; this is arguably a broader variation of neurodiversity, but similarly highlights natural human variation, as well as need for recognition and acceptance (Jaarsma & Welin, 2012). Within this category, people with Down syndrome and their family members express value for disability, as lives worth living and as mutually beneficial, and as productive; also, several of the advocacy organizations discuss inclusion as part of the organization's agenda. Particularly within the community outreach category, the social model of disability also surfaces. Depending upon the audience, some of the videos within this category might fit so-called "inspiration porn", in that typical activities are framed as extraordinary when someone with a disability performs them (Ellis, 2015). In this manner, the images of everyday life may be interpreted as the best expected outcome for someone with Down syndrome, rather than a typical outcome.

Language about abortion is used predominantly to report high percentages of selective termination following prenatal diagnosis of Down syndrome. While the videos do not condemn such pregnancy management decisions, the videos across all categories highlight stories about resisting this norm. More commonly, the videos connect such selective termination to eugenics; however, the relationship between the two is left somewhat ambiguous. Eugenics is articulated broadly, as the elimination of people with disabilities or undesirable traits, or as prevention of

lives deemed as not worth living. Most of these videos focus on the choice to have a child, regardless of testing results, suggesting a potential value in knowing about Down syndrome prenatally. Broadly, more information is discussed as inherently good, including more information about life and family life with disability.

The major limitation of this project relates to external validity. While this analysis included a representative sample of YouTube videos and YouTube has global reach, we only identified videos retrieved using the embedded search function feature and only included English language videos. I expect notable selection bias with regards to the people who post videos and a likely tendency for videos to express extreme viewpoints to increase viewership; this may suggest that some attitudes and ideologies are common or popular, when in actuality they are not. While there is some anonymity when posting, we also imagine that people may be more likely to post videos that they think others will find acceptable and appropriate. I make no claim that what was identified here is representative of public attitudes about prenatal testing for Down syndrome. However, the work does show at least some of the diversity of information circulating in the public sphere about such testing. Given the popularity of this video-sharing site, I take these publicly available narratives to represent some common understandings about prenatal testing for Down syndrome.

CONCLUSION

YouTube is a publicly available, easily accessible platform for information sharing. From the outset of this analysis, I expected personal narratives about prenatal testing experiences to be readily documented in this venue. Interestingly, almost none were retrieved. Instead, much of what was found embodied pro- information and pro-life ideologies. While some had religious undertones, the majority more explicitly highlight a desire to share information about

the value of living with Down syndrome and how such lives contribute meaningfully to the lives of their family members and friends. Of all the things that people might share about their experiences with prenatal testing, I am surprised how dominant this message is in these videos. Given my methodological approach, I cannot say with any certainty why people are thematically posting this message. I infer that many of the parents posting videos specifically about prenatal testing were likely offered and even accepted prenatal testing, I take this recurrent theme to mean they felt poorly informed or misinformed about life with Down syndrome. A large number of videos identified and analyzed here did not directly address prenatal testing specifically. In these instances, I think this theme might result as a reaction to negative or inaccurate assumptions circulating publicly about living with Down syndrome. People with Down syndrome and their families convey publically that the lives of people with Down syndrome are meaningful and valuable. In a couple of instances this seems to reflect a social model-oriented understanding of disability, where discriminatory social structures and practices are defined as the predominately issues for people with Down syndrome, as opposed to the biophysiological impacts of trisomy 21 (Oliver, 1990). More commonly, however, the videos seem to perpetuate attitudes of seemingly justified paternalism and pity. Again, I cannot determine if this is the intent or if it is simply an outcome of the content and framing. For the most part, the videos are not judgmental and do not impose that others should make a similar decision to continue pregnancy as these parents of children with Down syndrome have; instead, these parents are making their private lives public, so that others have a more holistic view about living with Down syndrome.

If we are to promote the use of YouTube videos to support value-consistent decision-making a prenatal testing and subsequent pregnancy choice, then there is need for quality assessment tools. We might imagine that such quality assessment tools should be created in a

collaborative fashion including community members, health educators and clinicians. Such tools should empower the public to evaluate YouTube videos for the health information they share and access, so that they can more easily assess if the content is accurate and up-to-date. With regards to videos about prenatal testing for Down syndrome, there is also need for broader representation, including more racial and ethnic diversity, as well as inclusion of videos posted by people who choose to terminate following prenatal diagnosis; in this manner, those facing decisions about prenatal testing and subsequent pregnancy choices will gain access to perspectives and experiences that may more closely align with their own that are marginally or unavailable in other sources. In addition, we might imagine YouTube videos might be an appropriate and useful place for increased public dialogue about genetic testing and its ethical, legal, and social implications, given the ease for which diverse range of opinions can be expressed and shared. Given the underrepresentation of people with Down syndrome in YouTube videos about prenatal testing, Disability Studies scholars and disability rights advocates should consider engaging in this form of media exchange, in order to provide prospective parents with an alternative perspective.

Given the potential opportunities for patient education, engagement and advocacy, health professionals might consider contributing to creation and development of YouTube videos. At the very least, YouTube videos might help to supplement paper-based health education materials, and this might help to ensure evidence-based health education is publicly available, easy to access, and appropriate for patients with low literacy levels. Inter-professional efforts and collaboration with advocacy organizations help to ensure that the best and most wanted information is available to patients seeking health information to better inform their personal decision-making about such tests.

References

ACOG Office of Communications. (2007a). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

ACOG Office of Communications. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm

Alderson, P. (2001). Down's syndrome: cost, quality and value of life. *Social Science & Medicine (1982), 53*(5), 627-638.

American College of Obstetricians and Gynecologists. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. *Obstetrics and gynecology, 110*(6), 1459.

American College of Obstetricians and Gynecologists. (2012). Noninvasive prenatal testing for fetal aneuploidy. *Obstet Gynecol, 120*(545), 1532–1534.

Americans with Disabilities Act, Pub. L. No. 110-325 (1990).

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.

Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Asch, A. (2003). Disability equality and prenatal testing: contradictory or compatible? *Florida State University Law Review. Florida State University. College Of Law, 30*(2), 315-342.

Baily, M. A., & Murray, T. H. (2008). Ethics, evidence, and cost in newborn screening. *Hastings Center Report, 38*(3), 23-31.

Bakke, A. (2013). Disabling the Rhetoric of Prenatal Genetic Testing: A Critical Analysis of Online Self-Help Literature. *Critical Disability Discourse, 5*, 11-38.

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? *The American Journal of Bioethics, 9*(8), 48-56.

Biesecker, B. B., & Hamby, L. (2000). What difference the disability community arguments should make for the delivery of prenatal genetic information. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.

Bindra, R., Heath, V., Liao, A., Spencer, K., & Nicolaides, K. H. (2002). One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. *Ultrasound in Obstetrics and Gynecology, 20*(3), 219-225. doi: 10.1046/j.1469-0705.2002.00808.x

Botkin, J. R., Clayton, E. W., Fost, N. C., Burke, W., Murray, T. H., Baily, M. A., . . . Ross, L. F. (2006). Newborn Screening Technology: Proceed With Caution. *Pediatrics, 117*(5), 1793-1799. doi: 10.1542/peds.2005-2547

Browner, C. H. (2000). Situating Women's Reproductive Activities. *American Anthropologist, 102*(4), 773-788.

Browner, C. H., & Press, N. (1996). The production of authoritative knowledge in American prenatal care. *Medical Anthropology Quarterly, 10*(2), 141-156.

Browner, C. H., & Press, N. A. (1995). The normalization of prenatal diagnostic screening. *Conceiving the new world order: The global politics of reproduction*, 307-322.

Buchanan, A., Brock, D. W., Daniels, N., & Wikler, D. (2000). *From Chance to Choice: Genetics and Justice*. Cambridge: Cambridge Univ Pr.

Burke, W., Atkins, D., Gwinn, M., Guttmacher, A., Haddow, J., Lau, J., . . . Wiesner, G. L. (2002). Genetic Test Evaluation: Information Needs of Clinicians, Policy Makers, and the Public. *American Journal of Epidemiology, 156*(4), 311-318. doi: 10.1093/aje/kwf055

Chao, S., Roberts, J. S., Marteau, T. M., Silliman, R., Cupples, L. A., & Green, R. C. (2008). Health behavior changes after genetic risk assessment for Alzheimer disease: The REVEAL Study. *Alzheimer disease and associated disorders, 22*(1).

Childress, J. F., Faden, R. R., Gaare, R. D., Gostin, L. O., Kahn, J., Bonnie, R. J., . . . Nieburg, P. (2002). Public health ethics: mapping the terrain. *The Journal of Law, Medicine & Ethics, 30*(2), 170-178.

Chou, W.-Y. S., Hunt, Y., Folkers, A., & Augustson, E. (2011). Cancer Survivorship in the Age of YouTube and Social Media: A Narrative Analysis. *J Med Internet Res, 13*(1), e7. doi: 10.2196/jmir.1569

Clerici, C. A., Veneroni, L., Bisogno, G., Trapuzzano, A., & Ferrari, A. (2012). Videos on rhabdomyosarcoma on YouTube: an example of the availability of information on pediatric tumors on the web. *Journal of pediatric hematology oncology, 34*(8), 329-331.

Constantin, C. M., Faucett, A., & Lubin, I. M. (2005). A primer on genetic testing. *Journal of Midwifery & Women's Health, 50*(3), 197-204.

Cushing, P., & Smith, T. (2009). Multinational Review of English Language Disability Studies Degrees and Courses. *Disability Studies Quarterly, 29*(3).

Davis, D. S. (2010). *Genetic dilemmas : reproductive technology, parental choices, and children's futures*. Oxford; New York: Oxford University Press.

Eddens, K. S., Kreuter, M. W., Morgan, J. C., Beatty, K. E., Jasim, S. A., Garibay, L., . . . Jupka, K. A. (2009). Disparities by race and ethnicity in cancer survivor stories available on the web. *J Med Internet Res, 11*(4), e50. doi: 10.2196/jmir.1163

Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. *American Journal of Obstetrics and Gynecology, 191*(3), 1044-1048.

Ellis, K. (2015). Disability and Spreadable Media: Access, Representation and Inspiration PornDisability in Popular Culture: Focusing Passion, Creating Community and Expressing Defiance (pp. 139-158): Ashgate Publishing Limited.

Farrell, R., Hawkins, A., Barragan, D., Hudgins, L., & Taylor, J. (2015). Knowledge, understanding and uptake of non-invasive prenatal testing among Latina women. *Prenatal diagnosis*, n/a-n/a. doi: 10.1002/pd.4599

Fox, S. (2012). Pew Internet: Health. Retrieved 24 January, 2013, from http://www.pewinternet.org/Commentary/2011/November/Pew-Internet-Health.aspx

Fox, S. (2013). Social media in medicine. *The Who, What, Where, When & Why of Health Care social media*. https://youtu.be/QbP9DtDjBRk: Albert Einstein College of Medicine.

Fox, S., & Duggan, M. (2013). Health online. *Pew Research Center's Internet and American life Project*.

Gabarron, E., Fernandez-Luque, L., Armayones, M., & Lau, A. Y. S. (2013). Identifying Measures Used for Assessing Quality of YouTube Videos with Patient Health Information: A Review of Current Literature. *Interactive Journal of Medical Research, 2*(1), e6. doi: 10.2196/ijmr.2465

Gee, J. P. (2010). *An introduction to discourse analysis : theory and method*. New York [etc.]: Routledge.
Ginsburg, F., & Rapp, R. (1999). Fetal reflections: Confessions of two feminist anthropologists as mutual informats. In L. M. Morgan & M. W. Michaels (Eds.), *Fetal subjects, feminist positions*. Philadelphia: University of Pennsylvania Press.
Green, R. C., Roberts, J. S., Cupples, L. A., Relkin, N. R., Whitehouse, P. J., Brown, T., . . . Group, R. S. (2009). Disclosure of APOE genotype for risk of Alzheimer's disease. *The New England journal of medicine, 361*(3), 245-254.
Groce, N. E. (1985). *Everyone here spoke sign language : hereditary deafness on Martha's Vineyard*. Cambridge, Mass.: Harvard University Press.
Haller, B. A. (2010). *Representing disability in an ableist world : essays on mass media*. Louisville, KY: The Advocado Press.
Harris, A., Kelly, S. E., & Wyatt, S. (2014). Autobiologies on YouTube: Narratives of Direct-to-Consumer Genetic Testing. *New Genet Soc, 33*(1), 60-78. doi: 10.1080/14636778.2014.884456
Hiatt, H. H. (1975). Protecting the medical commons: who is responsible? *The New England journal of medicine, 293*(5), 235-241. doi: 10.1056/nejm197507312930506
Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. *Qual Health Res, 15*(9), 1277-1288. doi: 10.1177/1049732305276687
Hughes, B., Joshi, I., & Wareham, J. (2008). Health 2.0 and Medicine 2.0: Tensions and Controversies in the Field. *J Med Internet Res, 10*(3), e23. doi: 10.2196/jmir.1056
Ivry, T. (2008). At the Back Stage of Prenatal Care: Japanese Ob-Gyns Negotiating Prenatal Diagnosis. *Medical Anthropology Quarterly, 20*(4), 441-468.
Jaarsma, P., & Welin, S. (2012). Autism as a natural human variation: Reflections on the claims of the neurodiversity movement. *Health Care Analysis, 20*(1), 20-30.
Kalfoglou, A., Suthers, K., Scott, J., & Hudson, K. (2004). Reproductive genetic testing: What America thinks. *Washington, DC: Genetics and Public Policy Center*.
Kellogg, G., Slattery, L., Hudgins, L., & Ormond, K. (2014). Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing. *Journal of genetic counseling, 23*(5), 805-813. doi: 10.1007/s10897-014-9694-7
Kelly, S., & Farrimond, H. (2012). Non-invasive prenatal genetic testing: a study of public attitudes. *Public Health Genomics, 15*(2), 73-81.
Khoury, M., McBride, C., Schully, S., Ioannidis, J. P., Feero, W. G., Janssens, A. C. J., . . . Cargill, M. (2009). The scientific foundation for personal genomics: recommendations from a National Institutes of Health–Centers for Disease Control and Prevention multidisciplinary workshop. *Genetics in Medicine, 11*(8), 559.
Khoury, M. J., Jones, K., & Grosse, S. D. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. *Genetics in Medicine, 8*(3), 191.
Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: conversations with my son. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.
Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. *Science translational medicine, 4*(137), 137ra176-137ra176.

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Shendure, J. (2012). Noninvasive whole-genome sequencing of a human fetus. *Sci Transl Med, 4*(137), 137ra176. doi: 10.1126/scitranslmed.3004323

Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". *AJOB Primary Research, 2*(2), 8-20.

Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing. *Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70*(1), 7-9.

LaRusse, S., Roberts, J. S., Marteau, T. M., Katzen, H., Linnenbringer, E. L., Barber, M., . . . Relkin, N. R. (2005). Genetic susceptibility testing versus family history-based risk assessment: Impact on perceived risk of Alzheimer disease. *Genetics in Medicine, 7*(1), 48-53.

Lawson, K. L., Carlson, K., & Shynkaruk, J. M. (2012). The Portrayal of Down Syndrome in Prenatal Screening Information Pamphlets. *J Obstet Gynaecol Can, 34*(8), 760-768.

Learman, L. A., Kuppermann, M., Gates, E., Nease, R. F., Gildengorin, V., & Washington, A. E. (2003). Social and familial context of prenatal genetic testing decisions: Are there racial/ethnic differences? *American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 119C*(1), 19-26. doi: 10.1002/ajmg.c.10004

LeGreco, M., & Tracy, S. J. (2009). Discourse tracing as qualitative practice. *Qualitative Inquiry, 15*(9), 1516-1543.

Leung, T. Y., Chan, L. W., Law, L. W., Sahota, D. S., Fung, T. Y., Leung, T. N., & Lau, T. K. (2009). First trimester combined screening for Trisomy 21 in Hong Kong: outcome of the first 10,000 cases. *The Journal of Maternal-Fetal & Neonatal Medicine, 22*(4), 300-304. doi: 10.1080/14767050802430842

Lewis, C., Silcock, C., & Chitty, L. S. (2013). Non-invasive prenatal testing for Down's syndrome: pregnant women's views and likely uptake. *Public Health Genomics, 16*(5), 223-232. doi: 10.1159/000353523

Lippman, A. (1999). Embodied knowledge and making sense of prenatal diagnosis. *Journal of genetic counseling, 8*(5), 255-274.

Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. *American Journal of Human Genetics, 51*(4), 936.

Lo, Y., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W., & Wainscoat, J. S. (1997). Presence of fetal DNA in maternal plasma and serum. *the lancet, 350*(9076), 485-487.

Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. *The American Journal of Human Genetics, 62*(4), 768-775.

Lupton, D. (2014). Critical Perspectives on Digital Health Technologies. *Sociology compass, 8*(12), 1344-1359.

Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: a commentary. *American journal of medical genetics. Part A, 155A*(8), 1777-1785.

Malone, F. D., Canick, J. A., Ball, R. H., Nyberg, D. A., Comstock, C. H., Bukowski, R., . . . D'Alton, M. E. (2005). First-Trimester or Second-Trimester Screening, or Both, for

Down's Syndrome. *New England Journal of Medicine, 353*(19), 2001-2011. doi: doi:10.1056/NEJMoa043693
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. *Prenatal diagnosis, 19*(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b
Markel, H. (1997). Scientific advances and social risks: historical perspectives of genetic screening programs for Sickle Cell Disease, Tay-Sachs Disease, neural tube defects and Down Syndrome, 1970-1997. *Medical and Public Health Law*. http://biotech.law.lsu.edu/research/fed/tfgt/appendix6.htm
Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. *Sociology of health & illness, 32*(1), 37-56.
Markens, S., Browner, C. H., & Press, N. A. (1999). Because of the risks': how US pregnant women account for refusing prenatal screening. *Social Science & Medicine, 49*(3), 359-369.
Marteau, T., Drake, H., & Bobrow, M. (1994). Counselling following diagnosis of a fetal abnormality: the differing approaches of obstetricians, clinical geneticists, and genetic nurses. *Journal of medical genetics, 31*(11), 864-867.
Marteau, T., Plenicar, M., & Kidd, J. (1993). Obstetricians presenting amniocentesis to pregnant women: practice observed. *Journal of Reproductive and Infant Psychology, 11*(1), 3-10.
Mazanderani, F., O'Neill, B., & Powell, J. (2013). "People power" or "pester power"? YouTube as a forum for the generation of evidence and patient advocacy. *Patient Educ Couns, 93*(3), 420-425. doi: http://dx.doi.org/10.1016/j.pec.2013.06.006
Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf
Miron-Shatz, T., Hanoch, Y., Graef, D., & Sagi, M. (2009). Presentation Format Affects Comprehension and Risk Assessment: The Case of Prenatal Screening. *Journal of Health Communication, 14*(5), 439-450. doi: 10.1080/10810730903032986
Moorhead, S. A., Hazlett, D. E., Harrison, L., Carroll, J. K., Irwin, A., & Hoving, C. (2013). A New Dimension of Health Care: Systematic Review of the Uses, Benefits, and Limitations of Social Media for Health Communication. *Journal of Medical Internet Research, 15*(4), e85. doi: 10.2196/jmir.1933
Muller, C., & Cameron, L. D. (2015). It's complicated – Factors predicting decisional conflict in prenatal diagnostic testing. *Health Expectations*, n/a-n/a. doi: 10.1111/hex.12363
Murkoff, H. E., & Mazel, S. (2008). *What to expect when you're expecting* (4th ed.). New York: Workman Pub.
Nadler, H. L. (1968). Antenatal detection of hereditary disorders. *Pediatrics, 42*(6), 912-918.
Naslund, J. A., Grande, S. W., Aschbrenner, K. A., & Elwyn, G. (2014). Naturally occurring peer support through social media: the experiences of individuals with severe mental illness using YouTube. *PLoS One, 9*(10), e110171. doi: 10.1371/journal.pone.0110171

National Coalition for Health Professional Education in Genetics, & National Society of Genetic Counselors. (2012). Prenatal Screening/Testing Options for Trisomy 21 and Other Fetal Aneuploidies. In N.-I. P. T. N. Factsheet (Ed.).
Nelson, J. L. (1998). The meaning of the act: reflections on the expressive force of reproductive decision making and policies. *Kennedy Institute Of Ethics Journal, 8*(2), 165-182.
Nicolaides, K. H. (2011). Screening for fetal aneuploidies at 11 to 13 weeks. *Prenatal diagnosis, 31*(1), 7-15. doi: 10.1002/pd.2637
Nicolaides, K. H., Spencer, K., Avgidou, K., Faiola, S., & Falcon, O. (2005). Multicenter study of first-trimester screening for trisomy 21 in 75 821 pregnancies: results and estimation of the potential impact of individual risk-orientated two-stage first-trimester screening. *Ultrasound in Obstetrics and Gynecology, 25*(3), 221-226. doi: 10.1002/uog.1860
Norton, M. E., Brar, H., Weiss, J., Karimi, A., Laurent, L. C., Caughey, A. B., . . . Song, K. (2012). Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18. *American Journal of Obstetrics and Gynecology, 207*(2), 137.e131-137.e138. doi: http://dx.doi.org/10.1016/j.ajog.2012.05.021
Oliver, M. (1990). *The individual and social models of disability*. Paper presented at the Joint Workshop of the Living Options Group and the Research Unit of the Royal College of Physicians.
Oliver, M. (1996). *Defining Impairment and Disability: Issues at Stake.* Paper presented at the Exploring the divide, University of Leeds.
Ormond, K. E., Gill, C. J., Semik, P., & Kirschner, K. L. (2003). Attitudes of health care trainees about genetics and disability: Issues of access, health care communication, and decision making. *Journal of Genetic Counseling, 12*(4), 333-349.
Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine, 13*(11), 913-920.
Pilnick, A. (2004). 'It's Just One of the Best Tests that We've Got at the Moment': the Presentation of Nuchal Translucency Screening for Fetal Abnormality in Pregnancy. *Discourse & Society, 15*(4), 451-465. doi: 10.1177/0957926504043710
Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. *Health Expectations, 11*(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x
Press, N., & Browner, C. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.
Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.
Press, N., Wilfond, B., Murray, M., & Burke, W. (2011). The power of knowledge: How carrier and prenatal screening altered the clinical goals of genetic testing. In W. Burke, K. A. Edwards, S. Goering, S. Holland & S. B. Trinidad (Eds.), *Achieving Justice in Genomic Translation: Re-Thinking the Pathway to Benefit: Re-Thinking the Pathway to Benefit*: Oxford University Press, USA.
Rapp, R. (1993). Amniocentesis in sociocultural perspective. *Journal of genetic counseling, 2*(3), 183-196.

Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), *Representations of motherhood*: Yale University Press.
Rapp, R. (1998). Refusing prenatal diagnosis: The meanings of bioscience in a multicultural world. *Science, technology & human values, 23*(1), 45-70.
Rapp, R. (1999). *Testing women, testing the fetus: The social impact of amniocentesis in America*.
Reed, K. (2009). 'It's them faulty genes again': women, men and the gendered nature of genetic responsibility in prenatal blood screening. *Sociology of health & illness, 31*(3), 343-359. doi: 10.1111/j.1467-9566.2008.01134.x
Riley, C. A. (2005). *Disability and the media: prescriptions for change*. Hanover, NH: University Press of New England.
Roberts, J. S., Barber, M., Brown, T. M., Cupples, L. A., Farrer, L. A., LaRusse, S. A., . . . Green, R. C. (2004). Who seeks genetic susceptibility testing for Alzheimer's disease? Findings from a multisite, randomized clinical trial. *Genetics in Medicine, 6*(4).
Rothman, B. K. (1994). The Tentative Pregnancy: Then and Now. In K. H. Rothenberg & E. J. Thomson (Eds.), *Women and prenatal testing : facing the challenges of genetic technology*. Columbus: Ohio State University Press.
Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), *The disability studies reader* (2nd ed., pp. xviii, 451 p.). New York: Routledge.
Secretary's Advisory Committee on Genetic Testing. (2000). Enhancing the oversight of genetic tests: recommendations of the SACGT. from http://oba.od.nih.gov/oba/sacgt/highlights/sacgt012700summ.pdf
Shapiro, J. P. (1993). *No pity : people with disabilities forging a new civil rights movement*. New York: Times Books.
Skotko, B. G. (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? *Archives of disease in childhood, 94*(11), 823-826.
Snyder, M. W., Simmons, L. E., Kitzman, J. O., Coe, B. P., Henson, J. M., Daza, R. M., . . . Gammill, H. S. Copy-number variation and false positive prenatal aneuploidy screening results. *New England Journal of Medicine*.
Spencer, K., Souter, V., Tul, N., Snijders, R., & Nicolaides, K. H. (1999). A screening program for trisomy 21 at 10–14 weeks using fetal nuchal translucency, maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A. *Ultrasound in Obstetrics and Gynecology, 13*(4), 231-237. doi: 10.1046/j.1469-0705.1999.13040231.x
Stapleton, H., Kirkham, M., & Thomas, G. (2002). Qualitative study of evidence based leaflets in maternity care. *BMJ : British Medical Journal, 324*(7338), 639-639.
Steinbock, B. (2000). Disability, Prenatal Testing, and Selective Abortion. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights*: Georgetown Univ Press.
Stern, A. M. (2005). Sterilized in the name of public health: race, immigration, and reproductive control in modern California. *American Journal of Public Health, 95*(7), 1128.
Syed-Abdul, S., Fernandez-Luque, L., Jian, W.-S., Li, Y.-C., Crain, S., Hsu, M.-H., . . . Liou, D.-M. (2013). Misleading Health-Related Information Promoted Through Video-Based Social Media: Anorexia on YouTube. *J Med Internet Res, 15*(2), e30. doi: 10.2196/jmir.2237

Task Force on Genetic Testing, Holtzman, N. A., & Watson, M. S. (1998). *Promoting safe and effective genetic testing in the United States : final report of the Task Force on Genetic Testing*. Baltimore: Johns Hopkins University Press.

Taylor, J. S. (2000). Of sonograms and baby prams: Prenatal diagnosis, pregnancy, and consumption. *Feminist Studies, 26*(2), 391-418.

Ten Great Public Health Achievements--United States, 1900-1999. (1999). *JAMA: The Journal of the American Medical Association, 281*(16), 1481-1481. doi: 10.1001/jama.281.16.1481

Thomas, K., Bourke, J., Girdler, S., Bebbington, A., Jacoby, P., & Leonard, H. (2011). Variation Over Time in Medical Conditions and Health Service Utilization of Children with Down Syndrome. *The Journal of Pediatrics, 158*(2), 194-200.e191. doi: http://dx.doi.org/10.1016/j.jpeds.2010.08.045

US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml

van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. *Patient Educ Couns, 63*(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007

Vanzwieten, M., Willems, D., Knegt, L., & Leschot, N. (2006). Communication with patients during the prenatal testing procedure: An explorative qualitative study. *Patient Education and Counseling, 63*(1-2), 161-168. doi: 10.1016/j.pec.2005.09.013

Wald, N. J., Huttly, W. J., & Hackshaw, A. K. (2003). Antenatal screening for Down's syndrome with the quadruple test. *The Lancet, 361*(9360), 835-836. doi: http://dx.doi.org/10.1016/S0140-6736(03)12680-3

Wald, N. J., Huttly, W. J., Murphy, K. W., K, A., Bestwick, J. P., & Rodeck, C. H. (2009). Antenatal screening for Down's syndrome using the Integrated test at two London hospitals. *Journal of Medical Screening, 16*(1), 7-10. doi: 10.1258/jms.2009.008094

Yang, M., Seo, J., Patel, A., & Sansgiry, S. S. (2012). Content Analysis of the Videos Featuring Prescription Drug Advertisements in Social Media: YouTube. *Drug Information Journal, 46*(6), 715-722. doi: 10.1177/0092861512462020

Zick, C. D., Mathews, C. J., Roberts, J. S., Cook-Deegan, R., Pokorski, R. J., & Green, R. C. (2005). Genetic Testing For Alzheimer's Disease And Its Impact On Insurance Purchasing Behavior. *Health Affairs, 24*(2), 483-490. doi: 10.1377/hlthaff.24.2.483

Zimmern, R. L., & Kroese, M. (2007). The evaluation of genetic tests. *Journal of Public Health, 29*(3), 246-250. doi: 10.1093/pubmed/fdm028

Newspapers, radio and YouTube videos create a conflicting narrative about Down syndrome births since the implementation of prenatal genetic testing: some suggest that there are more births of infants with Down syndrome, while others suggest fewer. "Numbers" or quantitative data plays an important role in the public discourse, because they are used to direct conversations in particular ways and to make specific claims about the population-level impact of prenatal testing, including but not limited to having relatively no impact, preventing disability, and/or having so-called eugenic implications.

This section consists of two parts. The first part briefly discusses the move towards universal screening for Down syndrome, as well as population trends delaying childbearing and regarding pregnancy management following a prenatal diagnosis of Down syndrome that may influence Down syndrome epidemiology. The second part investigates the existing epidemiological evidence about Down syndrome birth rate in the United States from the Centers for Disease Control and Prevention and other academic literature. Overall, my review shows notable variance across studies reporting Down syndrome birth prevalence, in part, due to differences in purpose, data sources, data collection methodologies, and analyses. Ultimately, I highlight the important distinction between incidence and birth prevalence –particularly in the context of prenatally diagnosable conditions– and then, illustrate how different methodological approaches complicate reporting about how prenatal testing for Down syndrome impacts the number of infants born with Down syndrome.

Chapter 6 DOWN SYNDROME BIRTH PREVALENCE

Amniocentesis to diagnose Down syndrome prenatally has been a part of prenatal care for nearing 50 years (Nadler, 1968). Since then, scientific, technological, and social norms and practices have shifted: who receives such testing, what conditions we test for, what we know about the conditions we identify, how we address them in clinical settings and beyond. As a relatively common medical procedure, facts about such testing readily circulate in radio, news, and online for both potential patients and providers. Are circulating facts consistent with current evidence?

The 2007 ACOG policy (American College of Obstetricians Gynecologists) recommends: first, based upon good and consistent scientific evidence, amniocentesis should not be offered before 15 weeks of pregnancy; second, based upon limited or inconsistent evidence, amniocentesis after 15 weeks is safe; third, based upon consensus and expert opinion,

invasive diagnostic testing for aneuploidy −including Down syndrome− should be available to all pregnant women. ACOG (2007) specifies that nondirective counseling does not require a patient's commit to terminate her pregnancy in the case of a prenatal diagnosis. This policy reiterates earlier guidelines about prenatal screening and testing that recommends all pregnant women who receive prenatal care at or before 20 weeks of pregnancy –regardless of age− be offered screening and invasive diagnostic testing (American College of Obstetricians and Gynecologists, 2007). These clinical practice guidelines acknowledge growing evidence suggesting maternal age alone is not the best predictor of Down syndrome. Assuming that this guideline has diffused into clinical practice, we can expect that more patients will be faced with the choice about whether or not to accept prenatal screening and diagnostic testing.

Between 1995 and 2002, women who were 35 or older accounted for 13% of total births in the United States. By 2003-2006 and 2007-2012, this had already shifted to 15%. Appendix B illustrates these trends and births with Down syndrome by maternal age (2007-2012). Natality data was downloaded, cleaned, and analyzed (National Center for Health Statistics, 1995-2002, 2003-2006, 2007-2012). This trend towards older child-bearing suggests that more patients will experience Down syndrome-affected pregnancies.

While not all patients will necessarily accept prenatal screening for Down syndrome, more patients might accept this screening, given blood tests and ultrasound might be done without clearly distinguishing its use for prenatal screening from other pregnancy-related tests. This will likely increase in the numbers of prenatally diagnosed cases of Down syndrome. For some, such a diagnosis may result in a decision to terminate the pregnancy. Yet, current evidence provides an incomplete understanding about how many patients choose this.

Based upon an often-cited systematic literature review (Mansfield, Hopfer, & Marteau, 1999), pregnancy termination rates following a prenatal diagnosis of Down Syndrome were 92% in aggregate across 10 studies, ranging from 71%-100% (CI: 62%-100%). Only 3 of these studies (73 terminations/77 pregnancies) were based upon US context (years of study: 1980-1988), representing 1.5% (77/5035) of the total study population. Given that health services and social supports vary across countries and might influence decision-making, cultural context may be important in interpreting these results. Notably, patients who decide to forego invasive, diagnostic testing are excluded from these analyses. This bias may inflate termination rates, given the denominator is smaller than all pregnant women or all who undergo prenatal screening. This relatively small sample size and older timeframe may have limited generalizability to today's general US population.

A more recent systematic review (Natoli, Ackerman, McDermott, & Edwards, 2012) examines 24 studies across a broader range of years (study years: 1972 to 2007) and analyzes only US data. In investigating whether variation existed based upon study population, the authors categorized their findings into 3 groups (statewide birth defect registries, single-institution studies, and post-karyotype analyses). The mean weighted termination rate was 67%, ranging from 61%-93%, for statewide birth defect registries (n=7); 85%, ranging from 60%-90%, for hospital-based studies (n=9); and 50%, ranging from (0%-100%), for post-karyotype analyses (n=8). Each category of data has potential benefits and pitfalls that might shape the interpretation of the weighted termination rate, providing a more complete understanding about termination patterns following prenatal diagnosis when taken together: statewide birth defect registries studies are arguably more representative and less subject to selection bias (e.g., serving large proportions of patients with particular socio-demographics, like older women, certain

racial/ethnic groups, those with referrals to specialty care) than the other data sources, but oftentimes in order to capture such a large dataset, they rely upon both active and passive methods for identifying terminations; single-institution studies are representative of a more defined community (i.e., a particular patient catchment area with usually more narrow demographics), and given only a single institution's involvement in data collection, such studies oftentimes have more uniform data collection methods to identify a pregnancy termination; post-karyotype analyses may provide a clearer understanding of pregnancy management decisions (i.e., more completely identifying the termination rate due to prenatal diagnosis), but are susceptible to selection bias (e.g., increases in patient uptake increases the denominator and patients self-selecting to pursue invasive prenatal testing may not represent pregnant women generally). Overall, the review highlights variation in terms of mean weighted termination rates following prenatal diagnosis of Down syndrome across data sources, which appears largely attributable to differences in study population and data collection strategies. These reviews focus on pregnancy termination as the main outcome, as opposed to other outcomes measures.

In contrast, we sought to investigate the Down syndrome incidence. Incidence is the number of new cases per the at-risk population during a specified time frame (Gordis, 2009). As such, the true Down syndrome incidence would be the number of cytogenetically confirmed cases of Down syndrome (trisomy 21) over all pregnancies. Unfortunately, both the numerator and denominator are relatively difficult to assess. While Down syndrome cases are relatively easy to identify and confirm using perinatal genetic testing and using clinical indications of live-born infants, cytogenetic confirmation may not be performed in instances of pregnancy loss before or in the absence of amniocentesis or chorionic villus sampling. As such, we can expect case detection to be less reliable among both spontaneous and elective terminations, as well as

stillborn infants with Down syndrome than with pregnancies carried to term and resulting in live-born infants with Down syndrome. Arguably, the at-risk population is even more difficult to accurately capture; as with the cases, unconfirmed pregnancies, pregnancy terminations and pregnancy losses make data collection both difficult and under-reported. The true Down syndrome incidence simply cannot be reliably assessed with the evidence that is available. Commonly, public health surveillance relies upon birth prevalence −the number of cases regardless of pregnancy outcome per the number of live-births− as a proxy for this true incidence (Mason, Kirby, Sever, & Langlois, 2005).

Birth prevalence is an important population-level indicator that traditionally in public health is used to inform health program planning and guide resource allocation for services. As such, we systematically review Down syndrome birth prevalence within the existing literature, in order to discuss the variance in findings across studies. We highlight how methodological approaches may account for some of this variance, and we suggest some opportunities for more uniform data collection and reporting.

METHODS

For this review, PubMed database was searched for peer-reviewed studies. The search was restricted to articles published in English between 1990 and 2014 about Down syndrome in humans. During this comprehensive review process, all study types and study designs were included. The specific terms in the overview of search methods are provided. This comprehensive approach identified 2,105 articles related to Down syndrome incidence or prevalence. Given the purpose of this review, this broad approach was used to ensure that potentially relevant articles were not excluded.

In order to identify the most relevant articles about the Down syndrome birth prevalence, a set of specific inclusion criteria were applied. The articles were categorized based upon these criteria, in order to limit the initial search to articles that specifically addressed incidence or birth prevalence of Down syndrome in the United States or where this could be calculated from study data. These specific criteria were applied:

1. The primary or secondary purpose of the article addressed incidence or birth prevalence of Down syndrome. Number of Down syndrome cases had to be reported independently of other primary disabling conditions.
2. Studies needed to provide a case definition and to specify the study population. Both the numerator and denominator had to reflect exact numbers, as opposed to population estimates.
3. Studies had to use primary data in their analysis. General literature reviews, editorials, consensus reports, or commentaries were excluded. Secondary analyses of previously reported data, including data analyses of the Centers for Disease Control and Prevention - funded National Birth Defect Monitoring System, were also excluded.
4. Studies had to include US-based data.

Upon reviewing the abstract, 1,993 articles were excluded for not meeting these criteria. Based upon the full-text of these remaining articles, we excluded an additional 66 articles for not meeting these criteria. Two independent reviewers extracted data from the full-text of 46 included articles. This comprised of general information, including the author, funding support, purpose, study design, geographic region; methodological information, including data source, sample demographics, case definition, study population; outcome-related information, including potential strengths and weaknesses, discussion about prenatal testing, main outcomes related to Down syndrome birth prevalence in the general population and subgroups. This information was compiled in a Google Form™ designed specifically for this project. Data was downloaded and analyzed. The reviewers reconciled any discrepancies in the collected information through discussion until consensus was reached.

Eighteen studies were excluded from the pooled Down syndrome birth prevalence. Studies were excluded for the following reasons: specific numerator and/or denominator

estimated or not provided (n=2), case ascertainment from prenatal diagnosis of at-risk pregnancies (n=2), multiple analyses utilizing the same data set (n=8), analyses reporting only pooled data (n=4), and analyses based solely on birth certificate data (n=2). In instances where multiple analyses were completed using the same data set, the most inclusive in terms of years was included. Ten studies reported data about terminated cases and were included in the sub-analysis.

FINDINGS

In total, twenty-eight articles were identified and included in the systematic review (Benn, Egan, Fang, & Smith-Bindman, 2004; Benn, Horne, Briganti, & Greenstein, 1995; Besser, Shin, Kucik, & Correa, 2007; Bishop, Huether, Torfs, Lorey, & Deddens, 1997; Bornstein *et al.*, 2009; Caruso, Westgate, & Holmes, 1998; Centers for Disease Control and Prevention, 1994; Cocchi *et al.*, 2010; Collins *et al.*, 2002; Dzurova & Pikhart, 2005; Egan *et al.*, 2004; Egan *et al.*, 2011; Forrester & Merz, 1999, 2002, 2003; Hahn & Shaw, 1993; Henry, Britt, & Evans, 2008; Huether, Haroldson, Ellis, & Ramsay, 1996; James, 1993; Krivchenia, Huether, Edmonds, May, & Guckenberger, 1993; McDermott & Johnson, 2011; Olsen, Cross, & Gensburg, 2003; Olsen, Cross, Gensburg, & Hughes, 1996; Orton, Rickard, & Miller, 2001; Shin *et al.*, 2009; Siffel, Correa, Cragan, & Alverson, 2004; Vendola *et al.*, 2010; Wilson, Chan, & Herbert, 1992), and each article is summarized in Table 13. The articles were published between 1992 and 2011, representing data from 1972-2006 and fifteen states (Arkansas, California, Colorado, Connecticut, Georgia, Hawaii, Iowa, Massachusetts, New York, North Carolina, Ohio, Oklahoma, South Carolina, Texas, and Utah) Twenty-eight studies employ a cross-sectional study design. The majority of studies (n=22) utilize data from population-based registries; of these, 20 analyze data associated with the National Birth Defects Prevention Network and 2 use

data associated with the National Center of Health Statistics. Only 6 studies use hospital- or laboratory-based data. More than half did not report demographic information about maternal age (n=12) or race (n=10).

Case ascertainment varied greatly across studies; specifically, differences arose in terms of cases identification strategies (active, passive, or a combination), data sources (medical records, birth certificate data, physician reporting, registry reporting), case definitions (full trisomy, mosaicism, translocations, clinical indications, or a combination) and inclusion criteria (prenatal diagnoses, postnatal diagnoses, cytogenetic analyses, live-born infants, fetal deaths, pregnancy terminations stillborn infants, or a combination). The majority of articles discuss that prenatal testing and diagnosis (n=23) may influence Down syndrome birth prevalence, but overall, this discussion is limited and does not indicate whether more testing and diagnosis increases or decreases the number of live-born infants with Down syndrome. Most studies use state-level live birth data for the total live-born infants (denominator), presumably from birth certificate or vital statistic data; however, there was often limited discussion about exactly how this data was derived. In addition, there was variable inclusion of fetal deaths and elective terminations in the denominator. Most sub-analyses reported adjusted rates for maternal age equal to or greater than thirty-five years and race/ethnicity.

Based upon eleven articles, the pooled Down syndrome birth prevalence among the general population is 12.6 per 10,000 live births (9,368 cases/7,436,069) (Benn et al., 1995; Bishop et al., 1997; Caruso et al., 1998; Collins et al., 2002; Dzurova & Pikhart, 2005; Forrester & Merz, 2003; Hahn & Shaw, 1993; Henry et al., 2008; Krivchenia et al., 1993; Vendola et al., 2010; Wilson et al., 1992).

Ten articles were included in the sub-analysis (Bishop et al., 1997; Caruso et al., 1998; Cocchi et al., 2010; Dzurova & Pikhart, 2005; Forrester & Merz, 1999, 2002; Krivchenia et al., 1993; McDermott & Johnson, 2011; Siffel et al., 2004; Wilson et al., 1992). In total, 1,502 terminated cases were reported and 4,601,112 live births, for a calculated 3.3 (range: 0.7-7.6) terminated cases per 10,000 live births. For this sub-sample, the mean live birth prevalence is 11.1 per 10,000 live births. Based upon this analysis, if cases had not been prenatally identified and terminated, then the live birth prevalence would be 14.4 cases per 10,000 live births (Figure 1).

Table 13. Summary of included articles

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
Benn, 1995	Cross-sectional	Connecticut	University of Connecticut Cytogenetic Laboratory June 1992 – January 1994 Hospital-based	8.6	74	DS cases identified from amniocentesis report, alpha fetal protein reports, ultrasound and follow-up with physician, as well as information from regional genetic consultations and cytogenetic analysis from live born children 20	All patients screened with third marker testing 11,434	Yes – screen-positive and screen-negative	Cases confirmed with physician follow-up	Some patients who screened positive for DS, did not receive amniocentesis 26% follow-up information is incomplete, so case detection may underrepresent total number of cases	Yes	17.5 per 10,000 live births
Benn, 2004 Mt. Zion Women's Health Clinical Research Center	Cohort	Farmington, Connecticut	University of Connecticut Health Center 1991 – 2002 Hospital-based; processes data for region	12.5 (1991) 21.7 (2002)	-	Prenatal and Postnatal diagnosis of DS Prenatal cases identified based on cytogenetic laboratory result from amniocentesis or CVS. 270 (1991-2002)	Referrals for invasive prenatal testing 18,057	Yes – prenatal diagnosis testing format	99% of cases had karyotype result Multiple demographic fields were assessed to exclude duplicates Assess abnormal results in pregnancies where fetal death occurred prior to prenatal diagnosis	Maternal age was estimated based on birth certificate data	Yes	149.5 per 10,000 prenatal diagnostic tests over 12 year study

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
Besser, 2007	Cross-sectional	Atlanta, Georgia	Metropolitan Atlanta Congenital Defects Program (Registry) 1979 - 2003 Population-based birth defect registry	-	-	DS cases identified using hospital records and lab results 949 (1979-2003)	Actual number not specified	No subgroup analysis reported for birth prevalence	Population-based data Included all live births using active ascertainment strategies DS was confirmed cytogenetically	Birth prevalence is the secondary aim of this study Aggregate data reported U.S. Census data used for live birth rate Excluded Latinos from numerator but not denominator	No	8.9 per 10,000 births (1979-1983) 11.6 per 10,000 births (1999-2003) 13.0 per 10,000 live births (2003) in metropolitan Atlanta Birth prevalence was slightly higher among whites than among blacks in the annual percentage increase in prevalence was significant among whites, but not in blacks
Bishop, 1997	Cross-sectional	California	California Birth Defects Monitoring Program 1989 – 1991 Population-based birth defect registry	10.6	-	DS cases were identified by hospital and genetic center records 998 live births 469 electively terminated / 531 prenatal diagnosis 15 continued pregnancies resulted in spontaneous pregnancy loss	Population within all counties that the Program monitors 880,361	Yes – racial/ethnicity, selected counties	99% of these cases were confirmed by cytogenetic analysis High level of ascertainment for live-births given multiple data sources	Some elective abortions not accounted for given passive ascertainment methods	Yes	12.3 per 10,000 live births 14.4 per 10,000 Latino live births 9.3 per 10,000 White live births 13.5 per 10,000 Asian live births 10.9 per 10,000 Black live births during 1990-1991
Bornstein, 2009	Cross-sectional	-	Genzyme Genetics mid-trimester	100	-	DS cases identified from amniocentesis	All amniocentesis performed where the sole indicator	No	Relatively large sample size	Retrospective analysis	Yes	92.1 per 10,000 amniocentesis test in mothers ≥ 35 years of age

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			amniocentesis database Laboratory-based			399	was advanced maternal age 43,303			Numerator does not include analysis for general population, personal or family history of chromosomal anomaly		
Caruso, 1998 Massachusetts Developmental Disabilities Fund; Merrell Dow Pharmaceuticals; The Easter Seals Research Program; The New England Regional Genetics Group; The Peabody Foundation	Cross-sectional	Boston, Massachusetts	Brigham and Women's Hospital and Boston Children's Hospital 1972 – 1974 and 1979 – 1994 Hospital-based	-	-	DS cases identified through active ascertainment among all infants and fetuses 265	159,928 live births 1,303 stillbirths after 20th week 2,600 elective terminations during second trimester 1,805 infants with other major conditions (subtracted from total) 162,026	Yes—maternal age	Delineate between live births, stillbirths and electively terminated pregnancy effected by DS	Active malformation surveillance program, Monday through Friday (late 1980s coverage added on Saturday or Sunday) As a major teaching hospital, patients may not receive all obstetric care at the same facility	Yes	16.4 per 10,000 live births, stillbirths and selective terminations
Center for Disease Control and Prevention, 1994	Cross-sectional	United States	17 State surveillance programs (Arizona, Arkansas, California, Colorado, Georgia, Hawaii, Illinois,	-	-	Colorado, Illinois, Kansas, Maryland, Missouri, Nebraska, New Jersey, New York, North Carolina and Virginia: DS cases	All live births among the 17 states	Yes – race/ethnicity	Population-based data	Passive ascertainment strategy	Yes	9.2 per 10,000 live births

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			Iowa, Kansas, Maryland, Missouri, Nebraska, New Jersey, New York, North Carolina, Virginia, and Washington) 1983 – 1990 Population-based			identified by medical reports. Arizona, Arkansas, California, Georgia, Hawaii, Iowa, and Washington: DS cases identified by abstractor review of medical and other health records. 7,190	7,820,278		Diverse, regional data			
***Cocchi, 2010** Savstaholm Foundation; Medical Faculty of Uppsala University; Swedish Research Council	Cross-sectional	Atlanta, Georgia	Metropolitan Atlanta Congenital Defects Program 1993 – 2004 Population-based	**14.9 (<35 years old)	-	DS cased identified by reports of DS to registry Not specified	All live births 46,248 (mean number of births)	-	Population-based data	Passive ascertainment strategy	Yes	12.7 per 10,000 live births (1993-2004) 4.0 per 10,000 terminations (1993-2004) 16.7 per 10,000 live birth and terminations
Collins, 2002 National Institutes of Mental Health; cooperative agreement from the Centers for Disease Control; The South Carolina Department of Disabilities and Special Needs;	Cross-sectional	South Carolina	Greenwood Genetics Center, Medical University of South Carolina and University of South Carolina January 1990 – December 1999	-	-	DS cased identified by cytogenetic laboratory results, excluding translocations and inversion 616	All live births and fetal deaths in the State of South Carolina from January 1990 – December 1999 540,130	No	Collected majority of cases in State	Incomplete results from one site from 1990 – 1991 Cases performed outside the State may not have been accounted for	Yes	9.8 per 10,000 live births and fetal deaths (1992) 13.4 per 10,000 live births and fetal deaths (1999)

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
South Carolina chapter March of Dimes			Hospital- and Laboratory-based									
***Dzurova, 2005** Fogarty program for Czech Republic postdoctoral scholars	Cross-sectional	California	California Birth Defect Monitoring Program January 1996 – December 1997 Population-based	-	-	DS cases identified by prenatal or postnatal diagnoses of live births 593	All live births in the State of California 516,745	Yes – infant sex, maternal education, maternal age, paternal age	Population-based data	Self-report patient data	Yes	11.5 per 10,000 live births
Egan, 2004	Cross-sectional	United States	National Center for Health Statistics 1989 – 2001 Population-based	-	-	DS cased identified by review of birth certificate 1,954 (1989) 1,802 (2001)	All live births by birth certificates 4,040,958 (1989) 4,031,291 (2001)	-	Population-based data	Birth certificate underreporting	Yes	5.9 per 10,000 live births (1989) 4.6 per 10,000 live births (2001)
Egan, 2011	Cross-sectional	United States	National Center for Health Statistics 1989 – 2006 Population-based	-	78.9	DS cases identified by birth certificate checkbox 32,720 (reported) 65,492 (actual)	All live births 72,613,424	Yes – race/ethnicity, marital status, maternal education and region	Population-based data	Passive ascertainment strategy DS birth certificate is underreported	Yes	9.0 per 10,000 live births
Forrester, 1999 Centers for Disease Control and Prevention; March of Dimes	Cross-sectional	Hawaii	Hawaii Birth Defects Program 1987 – 1996	50.7	34.4	DS cases identified by active ascertainment from registry 306	193,917 live births 14,255 fetal deaths	Yes – maternal age, race/ethnicity, county, urbanity,	Population-based Multiple sources of case	Cases reported multiple times	Yes	14.7 per 10,000 reported live births and fetal deaths

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
Birth Defects Foundation; The Queen Emma Foundation; George F Straub Trust; Pacific Southwest Regional Genetics Network; The Kamehameha Schools/Bishop estate			Population-based				110 elective terminations 208,282	prior pregnancy, fetal sex	ascertainment ensures that cases are not missed Assess multiple demographic factors that may influence case outcomes			
Forrester, 2002	Cross-sectional	Hawaii	Hawaii Birth Defects Program 1986 – 1997 Population-based	13.0	30.0	DS cases identified by active ascertainment from registry 363	229,584 live birth 16,605 fetal death 129 elective terminations 246,318	Yes – race/ethnicity, county and urbanity	Population-based	Cases reported multiple times	Yes	14.7 per 10,000 live births and fetal deaths 8.67 per 10,000 unadjusted live birth 12.59 per 10,000 adjusted live births
Forrester, 2003 Hawaii State Department of Health Children with Special Needs Branch; Centers for Disease Control and Prevention; Ronald McDonald's Childrens Charities, March	Cross-sectional	Hawaii	Hawaii Birth Defects Program, a population-based birth defects registry for the entire state of Hawaii 1986-2000	13.4	58.4	DS case identified by cytogenetic analysis regardless of pregnancy outcome 384	All live births in the State of Hawaii 258,350	Yes – race/ethnicity	Population-based data Ethnically and racially diverse population All pregnancy outcomes included	Small number of cases when distributed across subgroups	No	14.9 per 10,000 pregnancies

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
of Dimes Birth Defects Foundation, George F. Straub Trust, Queen Emma Foundation, Pacific Southwest Regional Genetics Network, Kamehameha Schools/Bishop Estate												
Hahn, 1993	Cross-sectional	California	California Birth Defect Monitoring Program 1983 – 1988 Population-based	-	-	DS cases identified by abstracts of infants with DS ICD-9 codes and positive cytogenetic reports or clinical characteristics 1,058 live birth 979 confirmed with positive cytogenetic results	All live births 1,028,636	Yes – maternal age	Cases were linked to California Vital Statistics	Incomplete and inconsistent ascertainment of cases	Yes	10.3 per 10,000 live births
Henry, 2008	Cross-sectional	Colorado	Department of Public Health and Environment Birth Defect Registry 1989 – 2005 Number of births including the number of babies	-	-	DS cases identified by medical record review 1158	All live births 1,087,698	Yes – maternal age, race	Population-based county level data	Only cases in live births confirmed through a medical records review by the staff of the CPPHE were included	Yes	12.0 per 10,000 live births 8.1 per 10,000 live births among older mothers ≥ 35 years of age 32.6 per 10,000 live births among younger mothers < 35 years of age

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			born with DS; this data is gathered at the county level from the Colorado Department of Public Health and Environment Birth Defect registry							Passive ascertainment of cases		
***Huether, 1996**	Cross-sectional	British Columbia, Canada Ohio, Georgia and California	Ohio: University of Cincinnati or directly from 16 cytogenetic laboratories, 2 birth defect monitoring programs, birth certificates 1970 – 1989 (SW Ohio) 1970 – 1983 (rest of Ohio) California data was obtained from that genetic laboratories and genetic disease branch of the California health authority, which collects cytogenetic data statewide 1983 – 1991	8.2	62.3	DS cases identified by CVS and amniocentesis 3,102 Live births 5,509 Total 8,611	Fetuses 50,817 Live births 3,660,707 Total 3,711,524	Yes – maternal age, race, gestational age	Utilizes multiple population-based data sources, reducing impact of sampling variation	Relatively small sample sizes of translocations and mosaic DS	Yes	23.2 per 10,000 CVS, amniocentesis and live births

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			Metropolitan Atlanta Congenital Defects Program 1970 – 1989									
James, 1993	Cross-sectional	Continental United States	Birth defects monitoring program 1970 – 1987 Population-based birth defect registry	-	-	State reported cases based on hospital newborn discharge database Number not specified	Live- and stillbirths 15,487,449	No	Large sample size from 1,985 hospitals in 1,094 counties	Aggregate prevalence provided	No	8.2 birth prevalence per 10,000 live- and stillbirths
Krivchenia, 1993 Interagency agreement between the Centers for Disease Control and Prevention and the Department of Biological Sciences	Cross-sectional	Southwest Ohio and Atlanta, Georgia	Southwest Ohio: Ohio Department of Health and Centers for Disease Control and Prevention 1970 – 1989 Population-based Georgia: Metropolitan Atlanta Congenital Defects Program 1970 – 1989	-	75.8	Ohio: DS cases identified by cytogenetic results, birth certificates, obstetric medical records and Ohio Department of Health 750 Georgia: DS cases identified by trained abstractors assessing hospital records, birth certificates and cytogenetic laboratory results 546	Annual live births from Ohio Department of Health 554,897 Georgia Human Resources 350,635	Yes - race	Population-based data Active ascertainment strategy	Ascertainment strategy is subjective and subject to false-positive and false-negative	Yes	Ohio: 13.5 per 10,000 live births 1.0 per 10,000 fetuses Georgia: 15.6 per 10,000 live births 2.9 per 10,000 fetuses

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			Population-based									
McDermott, 2011 Maternal-child Health Leadership in Neurodevelopmental and Related Disabilities (MCH-LEND)	Cross-sectional	Hawaii	Hawaii State Department of Health Birth Defects Program 1997 – 2005 Population-based	-	-	DS cases identified at birth 134 Total DS pregnancies 218	All live births in the State of Hawaii 158,790	Yes – maternal age, ethnicity	Population-based data Assessed DS across all pregnancy outcomes, including live births, fetal death (before and after 20 weeks) and terminations	Small sample size among ethnic subgroup analysis	Yes	8.4 per 10,000 live births 13.7 per 10,000 live births and fetal deaths 26.5 per 10,000 live births among older mothers ≥ 35 years of age 4.8 per 10,000 live births among younger mothers < 35 years of age
Olsen, 1996	Cross-sectional	New York	New York State Congenital Malformations Registry 1983 – 1992 Population-based	38.7	76.9	DS cases identified by ICD-9 and BPA codes reported to New York State Congenital Malformations Registry Not specified	Not specified	Yes – maternal age, race/ethnicity	Population-based data	Raw data not provided Passive ascertainment strategy	Yes	10.4 per 10,000 live births (1983-1992)
Olsen, 2003	Cross-sectional	New York	New York State Congenital Malformations Registry 1985 – 1997 Population-based	-	-	DS cases identified by ICD-9 and BPA codes reported to New York State Congenital Malformations Registry Not specified	Not specified	Yes – maternal age	Population-based data	Raw data not provided Passive ascertainment strategy	**Yes**	9.9 per 10,000 live births (1983 – 1997) 34.9 per 10,000 live births 1983 among older mothers ≥ 35 years of age

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
										Analysis only done for selected years		21.6 per 10,000 live births 1988 among older mothers ≥ 35 years of age 7.7 per 10,000 live births 1983 among younger mothers < 35 years of age 9.4 per 10,000 live births 1985 among younger mothers < 35 years of age 6.8 per 10,000 live births 1997 among younger mothers < 35 years of age
Orton, 2001	Cross-sectional	Colorado	Colorado Responds to Children with Special Needs, Colorado's Birth Defect Monitoring and Prevention Program 1989 – 1991 Population-based	-	-	DS cases identified from the Colorado Responds to Children Special Needs, born to a Colorado resident or diagnosed before their 3rd birthday. 198 Definite cases identified through medical record review or contact with physicians 151	All live births in the State of Colorado 159,974	No	Population-based data	Passive ascertainment of cases Limited to pregnancies of Colorado residents	No	9.4 per 10,000 live births

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
Shin, 2009	Cross-sectional	Arkansas, Georgia, California, Colorado, Iowa, North Carolina, New York, Oklahoma, Texas and Utah	Population-based birth defect programs Arkansas: 1993 – 2002 California (11 counties): 1983 – 2002 Colorado: 1989 – 2003 Georgia (5 central counties of metropolitan Atlanta): 1979 – 2003 Iowa: 1983 – 2003 New York (excluded New York City): 1983 – 2003 North Carolina: 1989 – 1993 1995 - 2003	-	-	DS identified by live-born infants when they were > or = to 20 weeks of gestational age, had a birth weight of > or = to 500g and had a DS diagnosis indicated by ICD-9-CM or BPA codes 6,580	All live births 5,579,947	Yes – maternal age, race, infant gender, congenital heart defects	Population-based data	Confirmed cases for some states but all Used variable methods of ascertainment (active, combination of active and passive, and passive)	No	11.8 per 10,000 live birth (1999-2003) 38.6 per 10,000 among older mothers >35 7.8 per 10,000 among younger mothers

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			Oklahoma: 1994 - 2003 Texas: 1994 – 2003 Utah: 1995 – 2003									
Siffel, 2004 Centers for Disease Control and Prevention; Association of Teachers of Preventative Medicine	Cross-sectional	Atlanta, Georgia	Metropolitan Atlanta Congenital Defects Program 1990 – 1999 Population-based	13.1	47.6	Cases were identified from hospital reporting and vital statistics. Abstractors confirmed from ICD-9 codes for full trisomy 21 in medical records at hospitals and genetic laboratories. Starting in 1994, perinatal office records were also included. 601 556 cytogenetically confirmed	All live births derived from the state of Georgia birth certificates 410,174	Yes – maternal age and data sources	Population-based data	Change in ascertainment strategies	Yes	12.8 cases per 10,000 live births (1990-1999) 8.4 per 10,000 live births (excluding pregnancy terminations) and 8.8 per 10,000 live births (including terminations) during 1990-1993 10.1 per 10,000 live births (excluding pregnancy terminations) and 15.3 per 10,000 live births (including terminations) during 1994-1999
Vendola, 2010	Cross-sectional	Texas	Texas Birth Defects Registry 1999 – 2003	-	-	DS cases identified by BPA codes in medical records of mothers residing in the State of Texas and linked to vital statistic data, excluding	All live births in the State of Texas 1,827,317	No	Population-based data Large and diverse sample	Incomplete vital statistics	Yes	11.2 per 10,000 live births 11.9 per 10,000 live births and fetal deaths

Author, year and source of support	Study design	Geographic Region	Data source(s)	Age (%)	White (%)	Case definition (numerator)	Study population (denominator)	Subgroup Analysis	Strengths	Weaknesses	Discuss prenatal testing?	Results (standardized for comparability to cases per 10,000 live births)
			Population-based			mosaic and unconfirmed cases 2,041 live births 139 fetal deaths						
Wilson, 1992 Maternal and Child Health Service, United States Public Health Sciences	Cross-sectional	Los Angeles, California	University of Southern California Medical Center (LAC-USC) 1974-1988 Hospital-based	7.8	13.0	DS diagnoses based on electronic medical records from cytogenetic laboratory logs 319 (Latino) 29 (non-Latino)	All live-births at LAC-USC Patient reported ethnicity of Mexican or Central American heritage was classified as Latino 188,802 (Latino) 29,038 (non-Latino)	Yes – ethnicity, maternal age	Potential cases were confirmed by comparing patient medical records with chart review within the Genetics Division 98% of DS had confirmatory chromosomal analysis	Limited number of non-Latino subgroups	Yes	16.0 per 10,000 over 15 years. 16.9 per 10,000 live Latino births 10.0 per 10,000 live non-Latino births 16.7 per 10,000 (Latino) when adjusted for maternal age 17.5 per 10,000 when corrected for prenatal diagnosis

Authors designated with an asterisk (*) published studies that included both US and non-US data; only US data is reported in the Table.
Age is maternal age at birth equal to or greater than 35 years, unless noted.
Dashes (-) represent data not reported in the article.

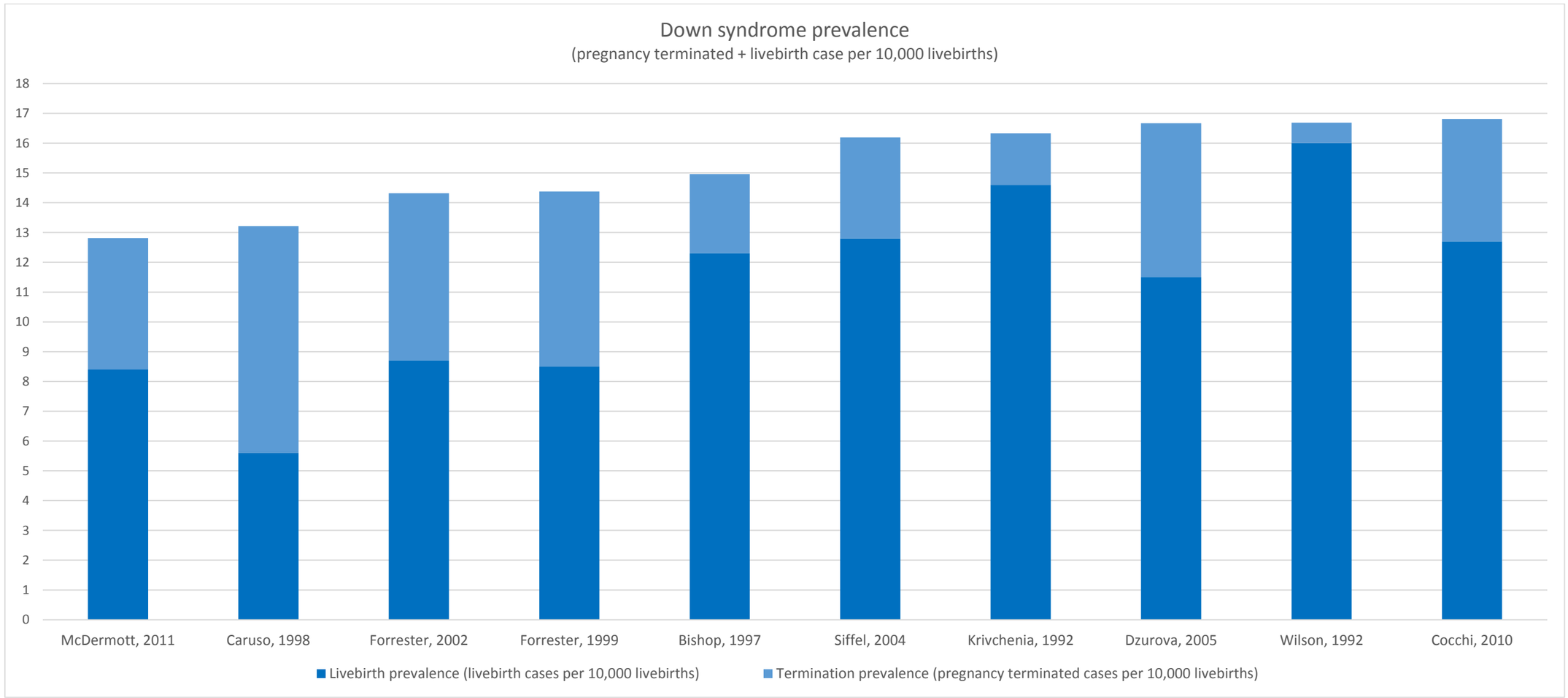


Figure 1. Cumulative Down syndrome prevalence

DISCUSSION

This review highlights notable variation in Down syndrome across years and geographic locations; however, we cannot completely assess the extent to which this variance results from actual differences. We expect at least some of the reported variance results from differences in methodological approach. Consistent with recommendations for birth defects surveillance (Correa-Villaseñor *et al.*, 2003; National Birth Defects Prevention Network, 2004), the majority of studies provide specific case definitions utilizing ICD-9 or British Pediatric Association codes and utilize trained medical specialists or medical records abstractors to identify cases. Relatively few sub-analyses are discussed. Perhaps because of concerns of privacy, most data is analyzed at a state level; county level, city level, and hospital level data is largely absent, even though such information is likely part of the data collection process. At the least, we should see sub-analyses for pregnant women equal to or greater than thirty-five years old and for race, but even these demographic categories are inconsistently reported (e.g., some states do not included maternal age on birth certificate data).

Six of the ten studies that reported terminated cases were based on data from either California or Hawaii. As such, we should generalize with caution. Hospital- or clinic-based data (2 studies) represent the extremes, 0.7 and 7.6 terminated cases per 10,000 live births. We might expect this variance to be attributed to differences in local tendencies, provider or hospital policies and practices, patient mix. This may suggest that hospital- and clinic-based data has greater variance than population-level data. Perhaps more importantly, however, this data suggests that at a population-level prenatal diagnosis of Down syndrome impacts birth prevalence, but fewer prenatally identified cases are terminated than we might expect based upon earlier research (Mansfield et al., 1999; Natoli et al., 2012).

Like other systematic reviews, the accuracy and reliability of the included studies limits this analysis. In addition, we restricted our search to studies published in peer-reviewed journals. Given that the National Birth Defect Prevention Network data sources provide useful and easy-to- analyze information about Down syndrome birth prevalence, this review is particularly susceptible to publication bias. Those with the resources to collect similar types of information may be reluctant to publish hospital- or laboratory-level findings, given the availability of population level data sources. As such, an even wider array of true variance may be masked. Similar to previous discussions about conceptual and methodological issues with systematic reviews and meta-analysis outside the context of birth defects surveillance (Saha, Chant, & McGrath, 2008), variation may itself be informative and larger samples may weigh the pooled prevalence.

Across studies there is wide variability with regards to case ascertainment that may bias the results and make compatibility difficult. For example, studies using birth certificate data, a passive case ascertainment strategy, comparatively under-report cases compared to other methodologies (Egan et al., 2004; Egan et al., 2011). In contrast, some studies use older women or those receiving diagnostic prenatal testing as their study population, which likely over-reports Down syndrome cases (Benn et al., 2004; Bornstein et al., 2009). While the majority of articles discuss prenatal testing as a factor that might influence case identification, in fact, most national data sets by and large do not collect sufficient data to distinguish prenatal and postnatal diagnoses. Given this heterogeneity and variance, the pooled birth prevalence should be interpreted with caution and should not be relied upon without consideration for variance.

Given these trends in the published literature, future research should make greater efforts to apply uniform case ascertainment strategies, so that comparability is possible; this will

minimize study bias that may create artificial variance in Down syndrome birth prevalence. In so doing, future research will be able to highlight true differences in Down syndrome birth prevalence across geographic regions that may be informative for health planning and programming. In addition to broadening the scope of existing evidence, such data may be used as a clinical indicator to inform hospitals and clinics. Given societal level changes in the demographics of pregnant women, collected information should include: maternal age, race/ethnicity, prenatal screening and testing utilized, time of diagnosis (if possible, week during pregnancy), and pregnancy outcome. If collected on an annual basis, such data would be more uniform across data sources and arguably more useful. Number of cases, case definitions, and reliable assessments of the study population (denominator) are needed to accurately inform both research and practice.

CONCLUSION

Because birth prevalence establishes the number of infants that Down syndrome impacts, it may be valuable to allocate adequate and appropriate resources for live-born infants with Down syndrome and their families, but we might question whether birth prevalence (i.e., the number of live-born infants with Down syndrome/total live births) is itself the best proxy measure to assess true incidence of Down syndrome. Reporting of Down syndrome birth prevalence often excludes pregnancy terminations, pregnancy losses, and still-born infants at least in the denominator and often some combination in the numerator. Incidence rate is more informative; particularly with regards to disease etiology, establishing the true incidence might help to identify risk factors associated with increases or decreases the number of fetuses affected with Down syndrome. As noted, the data currently available to assess true incidence is limited. Population-level data suggests minimal impact on Down syndrome birth prevalence due to

pregnancy termination following prenatal diagnosis of Down syndrome. The establishment of longitudinal, cohort studies of women as soon as they determine pregnancy may help to generate more complete data that is a better proxy for incidence than birth prevalence. Furthermore, such pregnancy registries might help to improve maternal and child health program planning, including perinatal health education and healthcare service coordination, like genetic counseling and pregnancy termination services.

Better data that investigates multiple measures across the reproductive decision-making process may help to ensure that appropriate, timely information is available to patients facing screening, testing and pregnancy management decisions, as well as the providers and policies supporting these decisions. Public health surveillance will be better equipped to address the health needs of our communities with more complete data about pregnancy termination rates, especially following prenatal diagnosis. Standardized, comparable measures across decision points might help individual hospitals and healthcare systems anticipate education and healthcare needs. We might expect that as our capacity to detect a wider range of condition increases and that more diagnoses will come with uncertain results, the ability to create processes and to have access to providers with the needed specialization to tackle patient questions will be important to providing high-quality prenatal care.

References

American College of Obstetricians and Gynecologists. (2007). Screening for Fetal Chromosomal Abnormalities. *Obstetrics & Gynecology, 109*(1).

American College of Obstetricians Gynecologists. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. *Obstetrics and gynecology, 110*(6), 1459.

Benn, P. A., Egan, J. F., Fang, M., & Smith-Bindman, R. (2004). Changes in the utilization of prenatal diagnosis. *Obstet Gynecol, 103*(6), 1255-1260. doi: 10.1097/01.aog.0000127008.14792.14

Benn, P. A., Horne, D., Briganti, S., & Greenstein, R. M. (1995). Prenatal diagnosis of diverse chromosome abnormalities in a population of patients identified by triple-marker testing as screen positive for Down syndrome. *Am J Obstet Gynecol, 173*(2), 496-501.

Besser, L. M., Shin, M., Kucik, J. E., & Correa, A. (2007). Prevalence of down syndrome among children and adolescents in metropolitan Atlanta. *Birth Defects Res A Clin Mol Teratol, 79*(11), 765-774. doi: 10.1002/bdra.20404

Bishop, J., Huether, C. A., Torfs, C., Lorey, F., & Deddens, J. (1997). Epidemiologic study of Down syndrome in a racially diverse California population, 1989-1991. *Am J Epidemiol, 145*(2), 134-147.

Bornstein, E., Lenchner, E., Donnenfeld, A., Barnhard, Y., Seubert, D., & Divon, M. Y. (2009). Advanced maternal age as a sole indication for genetic amniocentesis; risk-benefit analysis based on a large database reflecting the current common practice. *J Perinat Med, 37*(2), 99-102. doi: 10.1515/jpm.2009.032

Caruso, T. M., Westgate, M. N., & Holmes, L. B. (1998). Impact of prenatal screening on the birth status of fetuses with Down syndrome at an urban hospital, 1972-1994. *Genet Med, 1*(1), 22-28. doi: 10.1097/00125817-199811000-00006

Centers for Disease Control and Prevention. (1994). Down syndrome prevalence at birth--United States, 1983-1990. *MMWR Morb Mortal Wkly Rep, 43*(33), 617-622.

Cocchi, G., Gualdi, S., Bower, C., Halliday, J., Jonsson, B., Myrelid, A., . . . Anneren, G. (2010). International trends of Down syndrome 1993-2004: Births in relation to maternal age and terminations of pregnancies. *Birth Defects Res A Clin Mol Teratol, 88*(6), 474-479. doi: 10.1002/bdra.20666

Collins, J. S., Olson, R. L., DuPont, B. R., Wolff, D. J., Best, R. G., & Stevenson, R. E. (2002). Prevalence of aneuploidies in South Carolina in the 1990s. *Genet Med, 4*(3), 131-135. doi: 10.109700125817-200205000-00006

Correa-Villaseñor, A., Cragan, J., Kucik, J., O'Leary, L., Siffel, C., & Williams, L. (2003). The metropolitan Atlanta congenital defects program: 35 years of birth defects surveillance at the Centers for Disease Control and Prevention. *Birth Defects Research Part A: Clinical and Molecular Teratology, 67*(9), 617-624.

Dzurova, D., & Pikhart, H. (2005). Down syndrome, paternal age and education: comparison of California and the Czech Republic. *BMC Public Health, 5*, 69. doi: 10.1186/1471-2458-5-69

Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. *Am J Obstet Gynecol, 191*(3), 1044-1048. doi: 10.1016/j.ajog.2004.06.050

Egan, J. F., Smith, K., Timms, D., Bolnick, J. M., Campbell, W. A., & Benn, P. A. (2011). Demographic differences in Down syndrome livebirths in the US from 1989 to 2006. *Prenat Diagn, 31*(4), 389-394. doi: 10.1002/pd.2702

Forrester, M. B., & Merz, R. D. (1999). Prenatal diagnosis and elective termination of Down syndrome in a racially mixed population in Hawaii, 1987-1996. *Prenat Diagn, 19*(2), 136-141.

Forrester, M. B., & Merz, R. D. (2002). Epidemiology of Down syndrome (Trisomy 21), Hawaii, 1986-97. *Teratology, 65*(5), 207-212. doi: 10.1002/tera.10042

Forrester, M. B., & Merz, R. D. (2003). Maternal age-specific Down syndrome rates by maternal race/ethnicity, Hawaii, 1986-2000. *Birth Defects Res A Clin Mol Teratol, 67*(9), 625-629. doi: 10.1002/bdra.10112

Gordis, L. (2009). *Epidemiology*. Philadelphia: Elsevier/Saunders.

Hahn, J. A., & Shaw, G. M. (1993). Trends in Down's syndrome prevalence in California, 1983-1988. *Paediatr Perinat Epidemiol, 7*(4), 450-460.

Henry, G. P., Britt, D. W., & Evans, M. I. (2008). Screening advances and diagnostic choice: the problem of residual risk. *Fetal Diagn Ther, 23*(4), 308-315. doi: 10.1159/000123619

Huether, C. A., Haroldson, K., Ellis, P. M., & Ramsay, C. N. (1996). Impact of prenatal diagnosis on revised livebirth prevalence estimates of Down syndrome in the Lothian region of Scotland, 1978-1992. *Genet Epidemiol, 13*(4), 367-375. doi: 10.1002/(SICI)1098-2272(1996)13:4<367::AID-GEPI5>3.0.CO;2-1

James, L. M. (1993). Maps of birth defects occurrence in the U.S., Birth Defects Monitoring Program (BDMP)/CPHA, 1970-1987. *Teratology, 48*(6), 551-646. doi: 10.1002/tera.1420480605

Krivchenia, E., Huether, C. A., Edmonds, L. D., May, D. S., & Guckenberger, S. (1993). Comparative epidemiology of Down syndrome in two United States population, 1970-1989. *Am J Epidemiol, 137*(8), 815-828.

Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. *Prenatal diagnosis, 19*(9), 808-812.

Mason, C. A., Kirby, R. S., Sever, L. E., & Langlois, P. H. (2005). Prevalence is the preferred measure of frequency of birth defects. *Birth Defects Research Part A: Clinical and Molecular Teratology, 73*(10), 690-692.

McDermott, H., & Johnson, J. L. (2011). Is the prevalence of Down syndrome births in Hawai'i increasing? *Hawaii Med J, 70*(4), 72-76.

Nadler, H. L. (1968). Antenatal detection of hereditary disorders. *Pediatrics, 42*(6), 912-918.

National Birth Defects Prevention Network. (2004). *Guidelines for Conducting Birth Defects Surveillance* L. E. Sever (Ed.) Retrieved from http://www.nbdpn.org/docs/NBDPN_Guidelines2008.pdf

National Center for Health Statistics. (1995-2002). *Natality*. Retrieved from: http://wonder.cdc.gov/natality.html

National Center for Health Statistics. (2003-2006). *Natality*. Retrieved from: http://wonder.cdc.gov/natality.html

National Center for Health Statistics. (2007-2012). *Natality*. Retrieved from: http://wonder.cdc.gov/natality.html

Natoli, J. L., Ackerman, D. L., McDermott, S., & Edwards, J. G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). *Prenatal diagnosis, 32*(2), 142-153.

Olsen, C. L., Cross, P. K., & Gensburg, L. J. (2003). Down syndrome: interaction between culture, demography, and biology in determining the prevalence of a genetic trait. *Hum Biol, 75*(4), 503-520.

Olsen, C. L., Cross, P. K., Gensburg, L. J., & Hughes, J. P. (1996). The effects of prenatal diagnosis, population ageing, and changing fertility rates on the live birth prevalence of Down syndrome in New York State, 1983-1992. *Prenat Diagn, 16*(11), 991-1002. doi: 10.1002/(sici)1097-0223(199611)16:11<991::aid-pd977>3.0.co;2-5

Orton, H., Rickard, R., & Miller, L. (2001). Using active medical record review and capture-recapture methods to investigate the prevalence of Down Syndrome among live-born infants in Colorado. *Teratology, 64 Suppl 1*, S14-19. doi: 10.1002/tera.1079

Saha, S., Chant, D., & McGrath, J. (2008). Meta-analyses of the incidence and prevalence of schizophrenia: conceptual and methodological issues. *International Journal of Methods in Psychiatric Research, 17*(1), 55-61. doi: 10.1002/mpr.240

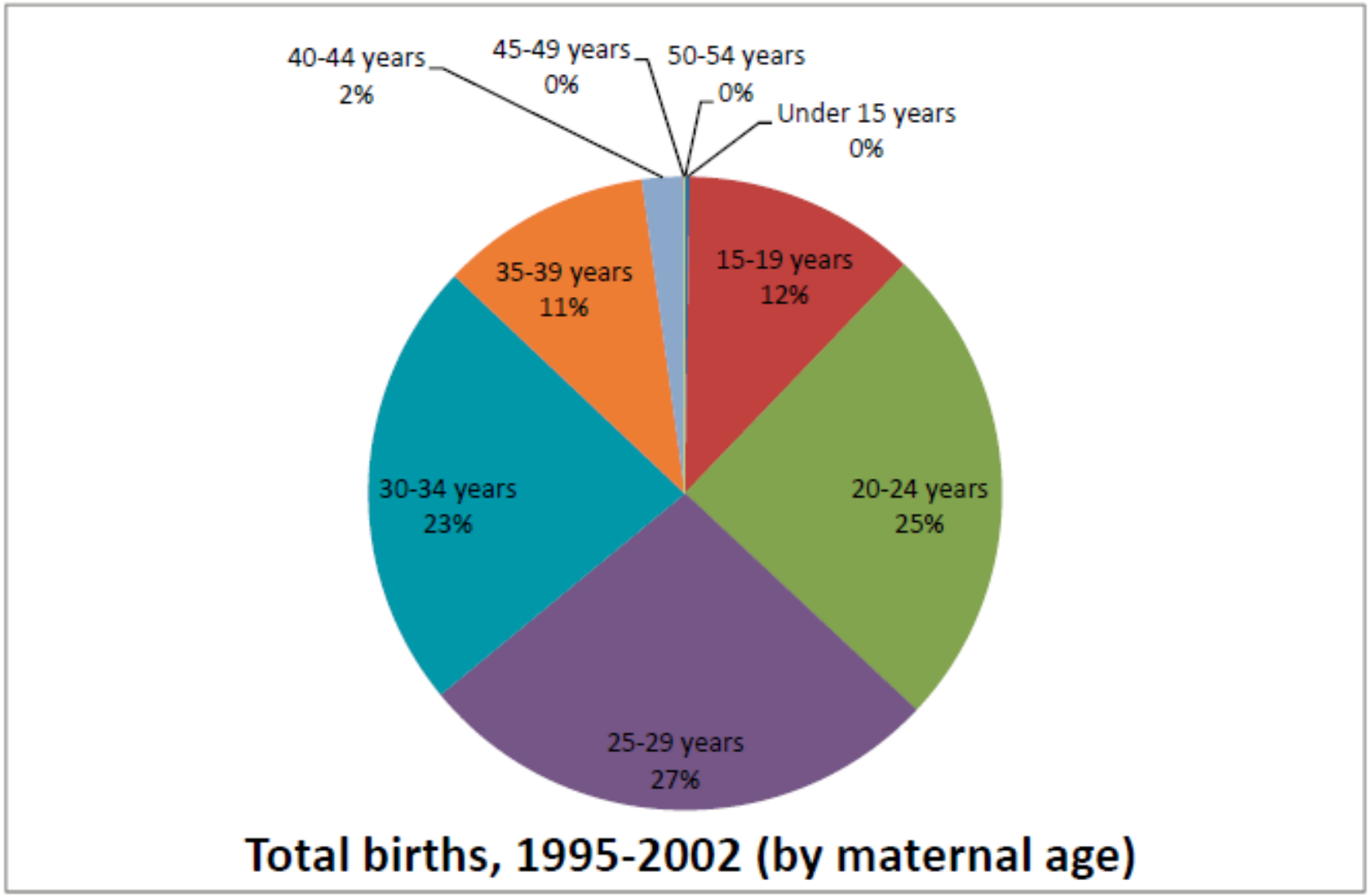
Shin, M., Besser, L. M., Kucik, J. E., Lu, C., Siffel, C., & Correa, A. (2009). Prevalence of Down syndrome among children and adolescents in 10 regions of the United States. *Pediatrics, 124*(6), 1565-1571. doi: 10.1542/peds.2009-0745

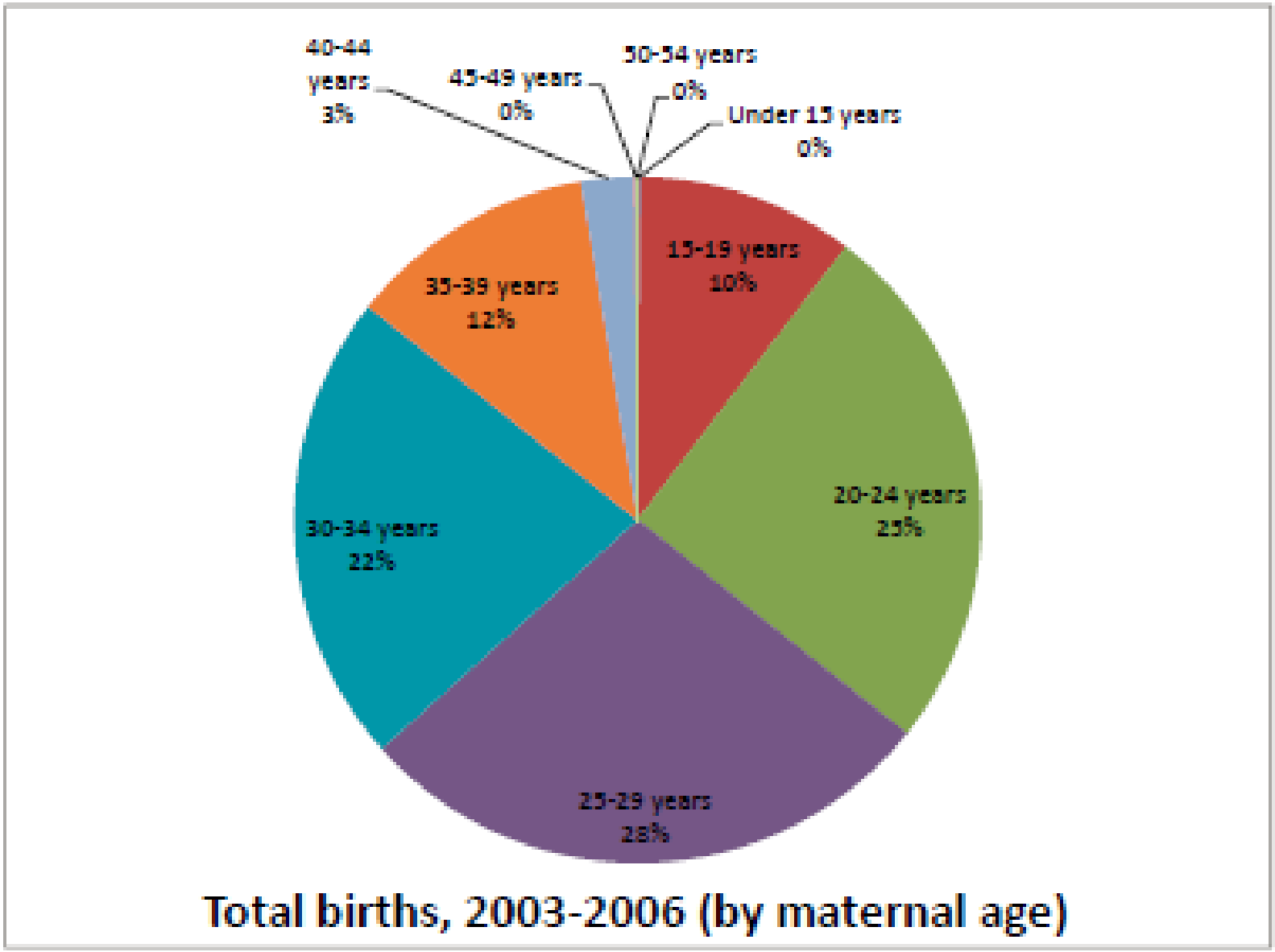
Siffel, C., Correa, A., Cragan, J., & Alverson, C. J. (2004). Prenatal diagnosis, pregnancy terminations and prevalence of Down syndrome in Atlanta. *Birth Defects Res A Clin Mol Teratol, 70*(9), 565-571. doi: 10.1002/bdra.20064

Vendola, C., Canfield, M., Daiger, S. P., Gambello, M., Hashmi, S. S., King, T., . . . Hecht, J. T. (2010). Survival of Texas infants born with trisomies 21, 18, and 13. *Am J Med Genet A, 152A*(2), 360-366. doi: 10.1002/ajmg.a.33156

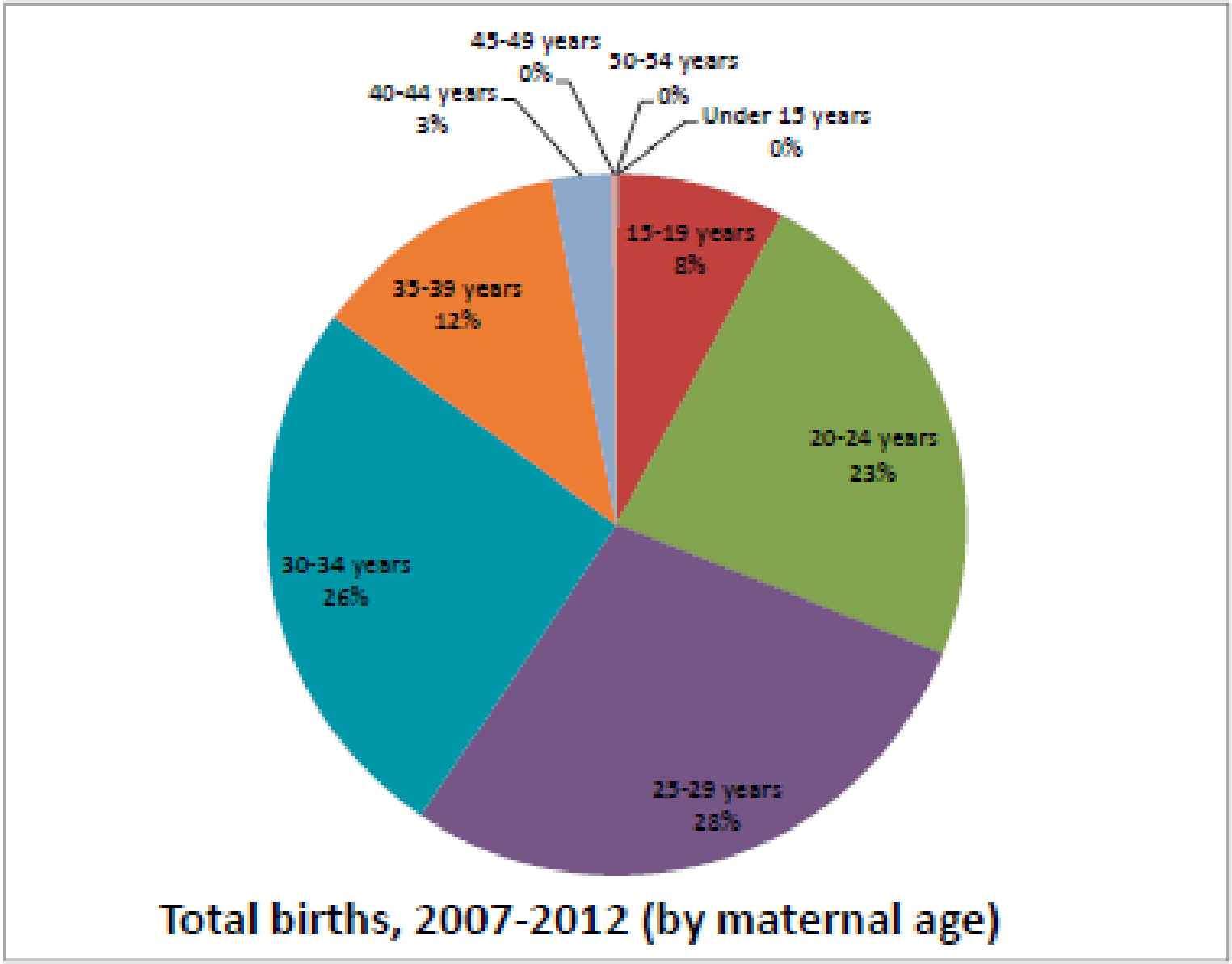
Wilson, M. G., Chan, L. S., & Herbert, W. S. (1992). Birth prevalence of Down syndrome in a predominantly Latino population: a 15-year study. *Teratology, 45*(3), 285-292. doi: 10.1002/tera.1420450307

Appendix B. Total births by maternal age





Total births, 2003-2006 (by maternal age)


45-49 years
0%
50-54 years
0%
40-44 years
3%
Under 15 years
0%
15-19 years
8%
35-39 years
12%
20-24 years
23%
30-34 years
26%
25-29 years
28%
Total births, 2007-2012 (by maternal age)

> This section provides a meta-synthesis of all the data that I analyzed for my dissertation. I provide an overview of all the data sources, major findings across these sources, and overall conclusions. Based upon these findings, I recommend strategies to improve health communication and health practice, as well as research, in this context.

Chapter 7 CONCLUSION

Nearly six years ago during an introductory course in public health genetics, I read my first article written by Adrienne Asch published in the American Journal Public Health (1999). At the outset of the article, it states:

> This article assumes a pro-choice perspective but suggests that unreflective uses of prenatal testing could diminish, rather than expand, women's choices. This critique challenges the view of disability that lies behind the social endorsement of such testing in the conviction that women will or should end their pregnancies, if they discover the fetus has a disabling trait (p. 1649).

This fit my own intuitions. As I read on and read more of her work, I felt as though Asch was not writing to me, as a public health professional; even though, she names public health professionals specifically, and her work is published in a prominent public health journal. To me, her attention focused first to obstetricians, midwives, nurses and genetic specialists involved with counseling prospective parents and second, to policymakers involved with the reproductive rights legislation. I could not identify what professional obligations she was suggesting for someone like me – a health educator -- despite my graduate-level education in health education, maternal and child health, and public health genetics. Looking back, this piece became a bit of a personal challenge, and led me to my first question: where does health education fit in conversations about genetics? I knew that I wanted to explore health communication and education within a broader social context −that is, outside and beyond the clinical encounter− for in this non-clinical environment, public health has a more substantive role.

Asch named primary care providers and genetic specialists as those primarily involved with counseling patients. Interestingly to me, these groups have distinct perspectives about

utility: primary care providers tend to adopt a more traditional view that focuses on clinically actionable results[12], while genetic specialists incorporate elements of personal utility. Public health professionals tend to understand utility from a population-level. Given this diversity, Asch's work led me to a second question: how do public texts create understandings about the clinical utility of prenatal testing? That is, how do encounters with health education outside the clinical setting shape ideas about the value and purpose of prenatal testing? ACOG shifted its policies about prenatal testing for Down syndrome and scientists have developed testing methods, I felt it was an interesting time to explore how language related to prenatal testing was evolving over time and to what extent it differed based on the source. I wanted a clearer understanding about the language used and its meaning, as a mechanism to better understand the underlying values, beliefs, and assumptions that drive healthcare decisions, in particular accepting or rejecting prenatal testing.

After witnessing much political rhetoric surrounding the 2008 presidential election, Republican Vice President Nominee Sarah Palin showcased her pro-life convictions by publicly sharing her experience of prenatal testing, Down syndrome diagnosis, and the choice to continue her pregnancy. I assumed that Disability Studies might have similar leanings towards this political ideology. As such, I found Asch's pro-choice, feminist, Disability Studies perspective intriguing. While I had been exposed to Disability Studies and worked in rehabilitation medicine before reading Asch's article, as I delved deeper into her work and into other feminist disability scholars, I have to admit that I was unconvinced that today's health messages −regardless of the source− were as negative, misinforming, and inaccurate about disability as Asch suggested. I

[12] In the Introduction (Chapter 1), a more complete description of the different views about clinical utility is described.

understood that Asch was making a philosophical argument about the moral permissibility of selective termination, but I was less interested in this. Instead, her work brought to my attention a third question: do today's health messages frame Down syndrome in a medically-oriented, negative light? If people with Down syndrome, their families, and friends emphasize the social barriers, as opposed to the medical ones, and discuss their lives positively, then how should health professionals craft health education and communication that more accurately reflect these sentiments? I think providing information about living with disability helps to ensure that patients are best and fully informed in decision-making about prenatal testing and subsequent pregnancy decisions.

Data and Analysis Overview

In efforts to address these questions, I explore health messages from a variety of sources –policy, radio, newspaper, biosciences literature, health education, and online videos (Table 14). In total, over 1,000 pages of text, two hours of audio, and five hours of audiovisual material were reviewed. I summarize the overarching findings by source in relationship to the primary research questions (Table 15), highlighting patterns in regards to involved stakeholders, constructed meanings of clinical utility, how cell free fetal DNA testing was described, the models of disability used to frame Down syndrome, the presence or absence of conversations about abortion and eugenics, as well as the portrayed values and expectations about choice in this context.

Table 14. Data catalogue

Data type	Data sources	Amount
Policy	American Congress of Obstetricians and Gynecologists; Disabled Persons International	~35 pages
Radio	NPR	140 minutes
	Public comments	475 posts
Newspaper	The New York Post; USA Today; The Spokesman-Review; The Philadelphia Inquirer; Los Angeles Times; The New York Times; The Denver Post; The Washington Post; The Baltimore Sun; St. Louis Post-Dispatch	~375 pages
Biosciences literature	Peer-reviewed journal articles about Down syndrome birth prevalence	112 (full-text)
Health Education	Web-based; print materials obtained directly from practitioners	~72 pages
Online videos	YouTube	53 videos

Table 15. Summary of findings

	Policy	Newspapers	Radio	Health Education	YouTube
Stakeholders	Obstetricians/Gynecologists; genetic counselors; Disabled People; pregnant women (patients)	Academic scholars; healthcare providers; industry consultants; individuals with expertise in ethical, legal, and social implications; people with Down syndrome; parents/family of children with Down syndrome; women as reproductive decision-makers; readers	NPR hosts, guests, commentators, listeners/viewers; White children with Down syndrome; Parents/family of children with Down syndrome; disability rights advocacy organizations; scientific experts/healthcare providers; people who chose to terminate after prenatal diagnosis; bioethicists	Prospective parents; genetic counselors; healthcare providers; March of Dimes; Center for Disease Control and Prevention; People with Down syndrome; parents/family of children with Down syndrome; disability rights advocacy organizations; healthcare organizations/system; private testing companies; major medical centers/systems	Disability rights advocacy organizations; healthcare providers; academic scholars; ethicist; religious leaders; legal advocates; pro-life and pro-choice; people with Down syndrome and their families; celebrities; genetic counselors

	Policy	Newspapers	Radio	Health Education	YouTube
Clinical Utility	ACOG: To gain knowledge to inform personal or healthcare decisions Focuses on personal utility, although hints to clinical utility by way of pregnancy termination DPI: To prevent lives judged to be abnormal; to prevent disease and impairment Focuses on public health utility	Financial implications of testing (insurance or out-of-pocket cost to consumer) To gain knowledge to inform personal or healthcare decisions Risk-benefit analysis Rarely uses language about utility, but largely uses language reflecting personal utility	To prevent disability; to gain personal knowledge; to gain knowledge which is conveyed as inherently valuable Conflates clinical and personal utility; suggests clinical utility by way of pregnancy termination	Too much information; confusing information No reference to clinical or personal utility specifically; instead, most outline potential risks and benefits	Too much or confusing information; more information is understood as inherently good, including information about life with disability No reference to clinical utility; little discussion of risks and benefits; general attention to broader ethical, legal, and social issues

	Policy	Newspapers	Radio	Health Education	YouTube
Cell free fetal DNA testing	N/A	Accurate; available during first trimester; more information; safer; timely results Too much or confusing information; emotional distress Eugenic Screening Concerns about accuracy; industry and regulatory involvement	Noninvasive; safer; more accurate; offered earlier in pregnancy	No risk of miscarriage; safer; most accurate screening; narrow range of conditions; ambiguous results in small percentage	New, noninvasive screening; low risk; may replace invasive methods Earlier detection; high sensitivity and specificity
Down Syndrome	ACOG: "genetic disorder"; "mental retardation" DPI: referenced once, but no details provided	Disability as a medical problem; "genetic disorder"; "mental retardation"	Disability as a medical problem; biomedical applications like prenatal testing offer the best means to address disability; in a few examples from comments, there are	Disability as a medical problem; "birth defect"; mental retardation;	Social model of disability; inspiration porn; disability as a medical problem; Down syndrome is a life worth living

	Policy	Newspapers	Radio	Health Education	YouTube
			references to diversity models of disability; "defects"; "abnormality"		
Abortion	ACOG: no explicit mention, but likely implicitly referred to by stating screening should be offered before 20th week of pregnancy DPI: disability as a legal exception for abortion; selective termination of undesirable traits	Women were framed as the sole decision-maker, particularly with regards to pregnancy termination Commonly reference statistic about 90% abortion rate following prenatal diagnosis of Down syndrome	Pregnancy termination is a mechanism to prevent disability and an appropriate course of treatment Based upon the comments, radio framing leads the audience to the conclusion that abortion is a treatment for Down syndrome	Largely avoided abortion especially in clinic-based health education materials, but fully acknowledged as a possible outcome and decision in health education materials produced by advocacy organization	Commonly references statistic about 90% abortion rate following prenatal diagnosis of Down syndrome
Eugenics	ACOG: no mention DPI: repeated use of "genetics" with language referencing issues with eugenics, like powerful agents oppressing less	Much on abortion and eugenics; direct connection between testing and pregnancy termination	Mentioned in public comments Several articles address history of eugenics, but little	No mention	Elimination of people with disabilities or undesirable traits; prevention lives

	Policy	Newspapers	Radio	Health Education	YouTube
	powerful ones, blurs the distinction between genetic and eugenics	conveyed as eugenic	sophistication or specification		deemed as not worth living
Choice	Expansion of choices ACOG builds upon understandings about healthcare consumerism, highlighting a wider array of options for patients. DPI aligns the term choice with bioethical notions about autonomy	Overlooks a potentially difficult decision making process Focus on selective termination following prenatal diagnosis	Readily expresses choice in relationship to pregnancy termination Testing results shape subsequent pregnancy choices, even though they do not cure, prevent disease, or yield sufficient knowledge to guide post-testing decisions public comments expand choice to discuss broader reproductive choices, including the ability to work with or around providers, the option of adoption, the option of in vitro fertilization	Provider-patient shared decision-making; informed consent Focus on personal choice of testing	Religious undertones; focus on choice to have a child regardless of testing results Emphasis on choice to have a child

Prenatal genetic testing for Down syndrome was represented in numerous ways. Across all sources, it was most often brought up as newsworthy by discussing existing methodologies, like quad screen or amniocentesis, in contrast to emerging ones, like cell-free fetal DNA testing. Many of the surges in media attention, particularly with regards to radio and newspaper coverage, happened around the time of publications in high impact journals that included discussion of prenatal testing (e.g., when Kitzman *et al.*, 2012 was published, several radiocasts and newspaper articles were published about it or including commentary from authors). Similarly, publications about policy changes or practice guidelines from notable professional organizations, like American College of Obstetricians and Gynecologists or the National Society of Genetic Counselors, also seemed to trigger media attention. Given that much of the publications and policy related to prenatal testing addresses cell-free fetal DNA testing, this methodology was a common discussion point in many of the texts. Representations of this methodology remained relatively constant over this time but had a common framing of novelty, timeliness, accuracy, and safety. Few sources specified that cell-free fetal DNA testing should be used only in pregnancies with high-risk for Down syndrome, given current evidence.

The stakeholders had notable diversity in terms of experience and expertise, but were similar across sources and time of publication or dissemination. Perhaps because of journalistic selection, the stakeholders represented a variety of viewpoints with varying degrees emphasis on particular values and concerns that were not necessarily thematically consistent by group (e.g., some healthcare providers expressed concern about the overwhelming amount of information, while others emphasized testing accuracy; people who had undergone prenatal testing expressed similar concerns as healthcare providers). Despite common reference to a high percentage of selective pregnancy termination, people who chose to terminate were left out of the public

conversation; there was only one significant contribution in the radio, where someone who had chosen to terminate following prenatal diagnosis was included as a commentator. This under-representation likely occurs because parents of children with Down syndrome who likely received prenatal testing are far easier to identify than those who choose to terminate, in part because a child with Down syndrome is a tangible, visible representation of a choice to continue a pregnancy. For those who choose to terminate equivalent representation of their choice does not exist. As such, researchers and journalist have to rely almost entirely upon self-report about selective termination following prenatal diagnosis, and for some, such disclosure may feel painful or stigmatizing, especially given the contentiousness surrounding abortion. This under-representation means that in research and in health messages the personal, psychosocial implications of selective termination following prenatal diagnosis are poorly documented. Therefore, those facing decisions about prenatal testing and diagnosis may have an incomplete understanding about the choice to terminate following such a diagnosis. While I find this lack of information alone problematic, I also think that this incomplete knowledge about the experience of terminating after a prenatal diagnosis perpetuates the notion that prenatal testing is a simple, routine procedure that responsible prospective parents ought to accept and fails to shed light on what for some is the chosen option of selective termination. Without clear, prominent examples, clinicians and health educators may feel discomfort speculating or beginning a conversation that is for most a deeply personal and value-laden decision.

Likewise, men involved with testing and subsequent pregnancy management decisions were also commonly left out, except in YouTube videos at lease to some extent fathers of children with Down syndrome illuminate their experiences. While parents of children with Down syndrome were often included in the conversation due to their experience with such

testing and reproductive decisions, their experience as parents of children with Down syndrome was not highlighted nearly as much. People with Down syndrome were rarely included, except in images of small children or discussion about childhood needs, including healthcare or education.

Most representations framed Down syndrome as a medical issue, stating several of the health consequences that people with Down syndrome commonly experience. The language used to describe Down syndrome is nearly identical across all sources, except YouTube videos, and times of publication or dissemination; in fact, it appeared to be stock language. From sources commonly generated by non-health professionals, selective pregnancy termination was readily discussed; however, in sources commonly generated by health professionals, like policy and health education, discussion about selective termination was largely omitted. Consistent with this pattern, texts that discussed selective pregnancy termination often mentioned eugenics as a potential societal impact, but texts that did not discuss selective pregnancy termination also did not mention eugenics. In addition, some texts that focused on the history of eugenics would sometimes refer to prenatal testing for Down syndrome as potential modern examples. The presence and absence, as well as the varying representations of eugenics, create a challenging space for any clear arguments or discussion for or against prenatal testing or selective termination. Because of the negative connotations associated with eugenics, its use in connection to prenatal testing tends to problematize particular choices or behaviors (i.e., selective termination). While this might be a deliberate usage, this language choice often focuses attention towards individuals making these decisions, rather than acknowledging the broader social circumstances that have constrained reproductive choices. For example, when individuals suggest that selective termination is eugenic, it suggests that selective termination is wrong, and

therefore, those choosing selective termination are doing something wrong. In so doing, this use of the term eugenics may overlooks how the social context, like a lack of adequate resources and supports for those parenting people with Down syndrome, may compel individuals to terminate following prenatal diagnosis.

Choices were discussed commonly across all sources, stakeholders, and times of publication or dissemination. However, the details about these choices varied across sources. Choice referred to patient choices within healthcare decision-making, particularly in reference to patient options within the healthcare system (e.g., provider influence on decision-making, financial costs and their influence on decision-making, and personal circumstances and their influence on decision-making) and to autonomy (i.e., informed consent, including the availability and knowledge about options, the ability to freely choose an option or alternative options). The use of choice was often situated in relationship to a larger conversation about reproductive choices, particularly focused on women's choices. Some of these choices included: utilizing in vitro fertilization or not; bearing a child or not; having a child at a particular time or not (e.g., delaying childbearing until established in a career); raising a nondisabled child or not; raising a child with a disability or not; accepting or rejecting prenatal screening; accepting or rejecting prenatal testing. Particularly by non-health professionals, choice was embedded in values about pro-life/pro-choice ideology; namely, the choice to terminate a pregnancy following prenatal diagnosis or the choice to raise a child with a disability with or without prenatal diagnosis. Yet, there is usually limited discussion about personal, financial, social circumstances that might shape these choices.

Clinical utility was rarely explicitly defined or described. Some texts implicitly illustrated the concept using two common frames. First, many texts implied that prenatal testing

offers early detection, and selective termination following prenatal diagnosis allows for disability prevention. Second, many texts implied that prenatal testing offers personal utility, providing valuable information that might shape personal and/or subsequent healthcare decisions. Too much lesser extent, some texts allude to: public health utility by referencing Down syndrome prevalence, cost-benefit or risk-benefit assessments by mentioning risk, benefits, and costs, by gaining information for its own value, and broader implications by referencing ethical, legal and social concerns. These meanings were commonly found across all sources to varying degrees and with varying levels of emphasis; additionally, the meanings largely did not change over this time period, despite cell-free fetal DNA translating from bench-to-bedside at the same time.

DISCUSSION

Taken together, these sources of health messages create a public discourse about prenatal testing for Down syndrome. Within this discourse, there are embedded values, beliefs and assumptions about cell-free fetal DNA testing, Down syndrome, abortion, eugenics and choice. I question whether any public consensus about the meaning of these terms exists, but the broad meanings combine and interconnect to shape how clinical utility is constructed and articulated. Because understandings about utility may shape decision-making, I contend that we need greater transparency about its meaning, both within and across disciplines. Different visions about clinical utility may led to different risk-benefit or cost-benefit assessments at a personal and policy level. I believe that this transparency will help to ensure that patients can access the information they need to make fully informed, pregnancy choices. While each chapter associated with a particular data source specifically addresses themes within and implications for health messages from source, here, I take a big picture view, discussing what these themes across health messages mean and matter. I believe this work has implications for most of the involved

stakeholders, and I provide some general guidance (Table 16). However, both at the outset and here at the conclusion I focus my attention towards those involved with health education and communication, professionally, outside the clinical encounter. Specifically, I address my original three questions. First, what do public messages say about genetics? Second, what understandings about the clinical utility of prenatal testing do public texts create? Third, how do today's health messages frame Down syndrome?

Table 16. General recommendations for stakeholders

Stakeholder	General Recommendations
People with disabilities, including people with Down syndrome and disability rights advocates	• Given the potential expansion of genetic knowledge regarding a wider array of genetic conditions or conditions with a genetic component, cross-disability coalition building may be important. • Make connections to journalists and healthcare providers involved with the offer of prenatal testing. At the very least, you might offer pictures and personal experience to their understanding or stories about disability. • Be mindful using terms like eugenics. If you choose to, explain what you mean, so that others understand your perspective; common perspectives from disability rights advocates often differ essentially from mainstream and clinician perspectives. • Asch and other feminist disability studies scholars created space and language about being pro-choice and questioning the moral permissibility of prenatal diagnosis and selective termination. Disability rights advocates should at the very least acknowledge that there is no singular Disability Studies perspective. Furthermore, within the disability community a diversity of perspectives are held about these issues, and a range of perspectives should be available in public statements and scholarship.
Journalists	• Investigate and report conflicts of interest. These conflicts are common among academically affiliated researchers and clinicians. • While air time and print can be expensive venues to provide all academic references to studies or study findings, many reports are available online and likely do not have similar constraints. By using web content or blogs, journalists may add important transparency to their work, which will better illustrate the connection to studies or study findings and better inform potential viewers. • Be cautious about accuracy when representing risk factors or risk. For example, some sources report that women over 35 have a greater risk of a Down syndrome-affected pregnancy, while women under 35 have a greater proportion of Down syndrome-affected pregnancies. While this statistic may be accurate in some contexts, its accuracy is dependent upon the number of pregnancies among women above and below 35. • For the most part, journalists do an exceptional job identifying diverse perspectives to include in their work about prenatal testing. More efforts could be made to include people of diverse racial and ethnic backgrounds; this may be best accomplished by contacting disability or Down syndrome-specific advocacy organizations.

Stakeholder	General Recommendations
	• While journalists often balanced in providing at least two perspectives, there is room to provide more balanced discussion about risks and benefits. Often times, mass media mentions benefits more extensively than risks, especially when reporting about scientific or medical developments.
Hospital and healthcare systems	• As more genetic tests become available, we might imagine that organizations develop programs connected to diversity and/or patient advocacy that can help support someone after decision-making. Because of the different yet condition-specific needs of particular families at different times, community members may be better resources than healthcare professionals with regards to advocacy. Their expertise regarding their lived experience makes them better suited to help others navigate healthcare decisions because of the similarity of their experience. For example, a family with a five-year-old with Down syndrome might assist someone with a newborn while receiving assistance from a parent of a high school student with Down syndrome. While some might argue that this should be done outside of the healthcare arena through social networking sites and/or support groups, I would argue that such a program demonstrates a health care institution's commitment to diverse patient groups - disability amongst other categories, like sex/gender/sexual orientation, race and ethnicity, education. We know that healthcare providers often do not have similar life experiences or shared characteristics with their patients as other patients might.
Obstetrician-gynecologists	• Dr. Google is not your competition, but she is the second opinion. Even a knowledgeable patient may have unanswered questions or lingering hesitancies about particular genetic tests. Many turn to the Internet as a resource. If someone enters a particular genetic condition, like Down syndrome, into a search engine, they return many results. The average user sifts through at least a few options. As my analysis suggests, the options related to Down syndrome reveals varied, sometimes seemingly conflicting, or incomplete information. In order to gain what they perceive as adequate information to make informed healthcare decisions, patients often visit a tremendous number of sites. Ask your patients about what information they are typing into their web searches; it might more accurately illustrate their fears, concerns and questions. Ask your patients about the sites they are visiting, because you might be able to steer them towards better sources. • Average primary healthcare providers likely have limited training in medical genetics, and as noted, they may not value nondirectiveness as much as medical genetics professionals. Therefore, I would argue that engaging an interdisciplinary team that includes a medical

Stakeholder	General Recommendations
	geneticists and/or genetic counselor is an important mechanism to ensure that patients have access to the resources needed to make informed decisions; we should make every attempt to minimize the need for a separate, additional clinic visit, if that is in the best interest of the patient. This approach might also include the development and housing of genetic-based health education materials among known maternal and child health entities, like March of Dimes and the centers for disease control and prevention websites. Publicly available health education materials will ensure that prospective parents have information about such testing, as well as potentially decision aid tools to guide them to information they may want, as well as questions they might want to ask healthcare providers. • The support and provision of public and clinical health education regarding preconception genetic counseling would create a less contentious space for medical genetics within reproductive decision-making. In part, this is simply echoing a call to better integrate genetic counseling within primary care settings. In the context of reproductive health, this likely means engaging mid-level professionals and paraprofessionals, like nurses, physician assistants, medical assistants, health educators, midwives and doulas. Given the already apparent demands on obstetricians/gynecologists and other primary care providers, better utilization of mid-level professionals might also help to create more opportunities for prospective parents to ask questions. • By giving patients the opportunity to engage with this information prior to the clinical encounter where decision-making needs to be made, they may feel more familiar with the implications of reproductive genetic testing and prepared to address decisions about testing, as well as its potential results. In this manner, we might better use limited patient-provider interaction time.

Stakeholder	General Recommendations
Genetic counselors	• For the most part, my analysis suggests clinic-based genetic counselors developed health education materials are often complete, of good quality, and reflect the diversity of perspectives. However, much of this information is behind closed doors, requiring access to primary care provider, a referral, and an additional visit for this specialty care. • Often times, educational materials are stored and located in personal drives or intranet servers rather than housed with public entities or community organizations. This is a stark contrast to most public health education, which in many ways focuses on dissemination in multiple arenas. In a similar regard, many maternal and child health websites offer more accessible information, but for the most part, lack comprehensive genetics education. I think the genetic counseling offers the genetic expertise, but public health has mastered community building and dissemination. • While genetic counselors are often connected to primary care providers via a referral mechanism, we need to create more interprofessional relationships between genetic counseling and public health. A typical relationship does not exist between genetic counseling and public health; therefore, both professions will have to be deliberate about making such connections. While this has implications for prenatal testing and maternal and child health, we might imagine similar collaborations should be developed in cancer detection and prevention programming. Working together, these partnerships to help to improve genetic literacy and ultimately, to serve as a foundation for a more informed public when faced with healthcare decisions related to genetic testing or other genetics related participation (like, screening, research, or even more broadly, navigating genetics representations in popular culture).

Stakeholder	General Recommendations
Bioethicists and researchers involved with ethical, legal and social issues of genetic testing	• This work provides some empirical support for Asch's normative claims. First, health messages − even outside the clinical encounter− portray conditions like Down syndrome extremely narrowly, focusing almost entirely on the medical implications. In so doing, health messages do not leave much opportunity to highlight that there is more to a person with such conditions than her medical state. Second, health messages perpetuate particular parental expectations. At least in the public sphere, these normative expectations strongly suggest a maternal responsibility towards a future child, including information- seeking and making informed decisions on behalf of a child. • There is decidedly space to further discuss the rights and responsibilities at-stake here: specifically, whose. In the public sphere, health messages focus almost complete attention on parental rights and responsibilities, extending almost no responsibility to society. • Particularly in this context, there is expressed concern about conflicts of interests. However, there is arguably insufficient guidance about what to do when such conflicts exist. Given growing private-public connections and biotech spinoffs from academia, disclosure itself is relatively meaningless. First, there needs to be more guidance about disclosures to whom. We might imagine that professional societies, including from journalism, science and health professional ones, be involved the creation and dissemination of such guidance
People facing decisions about prenatal testing and subsequent pregnancy management	• Before making pregnancy-related choices, we should reflect on our values and beliefs. • Choose clinical providers who you believe will provide you with accurate, up-to-date, unbiased information. Identify clinical providers who you believe will support your decision making. • We live in an era of information overload, so limiting the amount of time dedicated to information-seeking may actually be valuable. • We can access information from a variety of sources, but the sources may be of varying quality. Consult others and ask for help to assess good or bad sources. • The Internet is a great resource. As consumers, we often forget that search results are yielded in a non-objective way. In fact, I was - perhaps foolishly - shocked in doing my analysis how different the results were when I search using particular terms in comparison to colleagues, friends and family.

Based upon my methodological approach, I cannot assess whether journalists and others involved in health communication are directly accessing presumed newsworthy scientific developments from publications or whether they are accessing this information from a private company's, a professional organization's, or university's press releases. Following a publication and/or press release about such developments, news and radio sources proliferate human interest stories that incorporate bits of information from the publication and/or press release. For the most part, these stories engage multiple stakeholders and reflect multiple, often polarized, viewpoints about a particular related issue. The general story and framing remain similar, but the overall piece or portions can evolve and circulate in multiple editions and formats (e.g., publication to press release, press release to newsprint, newsprint weekday to weekend editions, etc.). As such, the life of a particular bit of information can have a relatively long lifecycle and viewership, as it circulates from one source to the next or from one medium to another. Yet, it does not appear that additional fact checking or substantive language changes are incorporated. For a public audience member –but even for me with access to additional knowledge and resources– this information does not have sufficient signposts or references needed to verify the information, as the recurrent un-cited statistic about termination rates of Down syndrome-affected pregnancies following prenatal diagnosis illustrates.

In general, cell- free fetal DNA testing provides an example of a recent development in scientific knowledge and as such, has received this mass media attention. The health messages highlight that cell-free fetal DNA testing is novel, timely, accurate and safe. While all of these features might be true, most sources provide insufficient information for me and likely for a public audience member to assess cell-free fetal DNA testing for these features ourselves. To the extent that prospective patients or their providers have read or followed news about such testing,

we might imagine that the focused attention on these features may make people more inclined to utilize such tests without adequate health education and communication within the clinical encounter. Some might argue that within the clinical encounter patients should have balanced information about testing options, this emphasis in the public sphere on the benefits of testing might suggest a need for more deliberate discussion about potential risks. I am definitely not saying that we should only discuss risks or benefits; however, in order to ensure that patients are fully informed about the prenatal tests available, I think we need to assess patient knowledge pretest and determine whether patients understand both the risks and benefits fully, including the potential out-of-pocket costs. In this manner, health education and communication will better address risk-benefit and cost-benefit assessments, which is expressed as a possible understanding of utility in the public sphere.

For the most part, health messages frame Down syndrome as an individual, medical issue. Furthermore, these messages accurately portray some of the potential health consequences associated with trisomy 21. This focus on particular medical issues without much of any discussion on potentially beneficial characteristics about people with Down syndrome may provide an incomplete portrayal about living with the condition, associated quality-of-life, and life for family members of someone with Down syndrome. To the extent that prospective parents engage with mass media, we might imagine that the emphasis on arguably negative health consequences may make people more inclined to utilize such tests, especially without alternative information. Given the dominant, negative frames of Down syndrome −even referring to the condition− may itself be directive.

Again, some might argue that within the clinical encounter patients should have evidence-based, accurate, up-to-date information about living with Down syndrome and its

implications for family life; however, to varying degrees, each of these features is absent from health messages broadly. First, this likely requires further research to establish a sufficient evidence about living with Down syndrome and its implications for family life. Second, many of the statements about health consequences associated with trisomy 21 do not indicate, even in approximations, how many people with Down syndrome experience particular conditions. For example, heart conditions may be associated with Down syndrome; however, few of the health messages indicate whether all people with Down syndrome experience this or a particular percentage of people with Down syndrome experience this[13]. Third, few health messages indicate when the evidence was established for claims about associated health conditions or quality of life. When health messages do not disclose what evidence supports its claims, then it is nearly impossible to confirm, refute, or know if one should question the information asserted. This is a major issue with the health messages circulating about prenatal testing for Down syndrome, as the statistic about 90% termination rates following prenatal diagnosis illustrates[14]. We might imagine that with improved medical intervention and greater inclusion in classrooms, workplace, and community environments that people with Down syndrome, like the general population, can live longer, healthier lives. Given the lack of longitudinal, population-level U.S. studies currently available we have insufficient evidence upon which to justify most claims about life with disability, family life with disability, birth prevalence of prenatally diagnosable genetic conditions. More research and investigation into these topics may help to improve decision

[13] To the best of my knowledge, a recent U.S. population-based, longitudinal studies exploring health conditions of people with Down syndrome does not exist. In a population-based Australian study, 95/208 (45.7%) had a cardiac issue or varying severity (Thomas *et al.*, 2011).
[14]I provide a more detailed description of this issue in newspaper articles (Chapter 5) and explanation about how quantitative data might not be representative of population-level trends (Chapter 7).

making that reflect up-to-date, unbiased, accurate knowledge. Evidence-based health education materials will empower patients to base their decisions off the best available information and improve the overall quality of information currently available in health messages.

In order to ensure that prospective parents have the best information available to them in their decision-making, I think health education and communication needs to better represent living with Down syndrome and its implications for family life across the lifespan. In this manner, health education and communication will better address personal utility of such testing; patients will have access to information they may perceive is valuable, in order to inform future decision-making. This notion of personal utility is the most commonly expressed notion of utility in the public sphere, so we might imagine that it is especially important to patients. The emphasis from YouTube videos, which in my analysis are arguably the best representation of public opinion, is that life with Down syndrome is valuable and contributes positively to family life; this common narrative from people who likely were at least offered prenatal testing suggests that this information is wanted and difficult to access or unavailable in mainstream health messages or even from within the clinical encounter. While as a whole, YouTube videos present a medical model of disability, including many instances of "inspiration porn", this platform offers the only counter-narrative to the medical model.

In popular health messages, Down syndrome is commonly connected to pregnancy termination, and in doing so, stakeholders create clinical utility by means of early detection using prenatal testing and disability prevention by means of selective termination following prenatal diagnosis. Given this understanding, some might argue that this is consistent with a traditional understanding of clinical utility; in this instance, prenatal testing leads to preventative healthcare services, albeit pregnancy termination. Alternatively, pregnancy termination following prenatal

diagnosis might also be understood as a tool for public health utility, in that early detection and disability prevention has population-level impact on Down syndrome birth prevalence. These applied understandings of clinical utility illustrate how different people might understand the offer of prenatal testing, its value, and normative understandings about what to do following prenatal diagnosis. If patients understand prenatal testing and selective termination as improving early detection and disability prevention or as a positive behavior that improves health outcomes, then they might feel expectation or obligation to terminate a pregnancy following prenatal diagnosis. I do not consider these perspectives to be inherently problematic, but rather different ways of understanding prenatal testing and selective termination. To me, what is problematic is that −especially in health education materials− this popular notion about prenatal testing's purpose (i.e., to detect Down syndrome early and prevent it) is largely unaddressed.

While health education materials do emphasize personal choice, few explicitly acknowledge pregnancy termination as a potential outcome following prenatal diagnosis. As such, patients may not completely understand that choosing screening may lead to a series of tests and potentially a diagnosis, which for some may alter their intended course of pregnancy. I assume that within the clinical encounter conversations about pregnancy termination of potential outcome are discussed, but my concern is that health education does not address this point well. To me, health education materials should address this, in part to improve comprehension about the testing process and in part to ensure that patients have access to important information without perceived time constraints and potential provider influence −intentional or unintentional. For some patients, more information, earlier about the potential outcomes of testing might result in different choices about screening and/or testing. I think this is especially true given the current availability of multiple, sometimes concurrent methods of screening. For example,

depending on timing, some might choose to forgo additional cell-free fetal DNA testing with known false positive rates in lieu of diagnostic testing despite its invasiveness. By incorporating more complete information about potential testing outcomes, health messages may better promote decision-making consistent with patient values. While there may be concern about raising issues related to pregnancy termination, particularly in the public sphere, mass media already engages discussion about termination following prenatal diagnosis. If publicly available health education materials and patient-provider conversations do not readily address pregnancy termination, then patients who seek health information outside of the clinical encounter may feel deceived or misled. In efforts to prevent these negative feelings, health education and policy guidance should directly mention pregnancy termination. Maintaining existing language about personal choice in conjunction with this is reasonable and appropriate, especially given this is such a publicly contentious topic. At the very least, I think that health education ought to say that in choosing screening patients may be faced with results that lead to more tests, including diagnostic ones, and that diagnostic test results may put some patients in the position of having to choose whether to continue or terminate the pregnancy. I think that better health education might help patients identify their values, as well as improve their understanding about the decision-making pathways. To me, presenting a few scenarios for patients to consider before making a decision about testing may be helpful (e.g., Are there any circumstances that you would consider termination? Let's imagine that someone's screening test comes back "positive". What does that mean? What might they do with that information? What options do you have now? What options do you have after screening? What options do you have after diagnostic testing?).

By connecting prenatal testing and selective termination, several stakeholders at different points raise concern about eugenics. While some mention specific historical instances of eugenics, eugenics is not defined nor explained. Instead, a broad and general references suggest prenatal testing is eugenic and that this is problematic. More so than other stakeholders, disability advocates identify key features about eugenics that they find problematic. Namely, the disability community highlights that selective termination prevents lives deemed as not worth living. Few advocates identify, specifically, who or how some lives are deemed not worth living, but the disability community does suggest that disability is commonly understood as an undesirable trait. The disability community offers a counter-narrative, suggesting that living with a disability is a valuable way of being in the world and that having a disability itself does not contribute negatively towards everyday life. Public reference to eugenics is one of the ways in which the public narrative acknowledges broader societal pressures and contextual issues associated with this testing. Far more commonly than concerns about eugenics, many stakeholders express assessing, understanding, and negotiating conflicting information or personal values is a central issue. Given the emphasis in public health messages on people who accepted prenatal testing, received a prenatal diagnosis, and chose to continue pregnancy, concern about adequate resources for people with Down syndrome is also mentioned. In these ways, the public narrative highlights an understanding of clinical utility that incorporates ethical, legal, and social issues.

I think we have to take the presence of ethical, legal, and social issues in the public narrative as an indicator that at least some people might recognize broader issues as part of their understanding of clinical utility. If people hold this understanding of clinical utility, then should patients be offered additional information about potential ethical, legal, and social issues of

prenatal testing? More so than evidence-based information or details about potential outcomes, I have to wonder whether this additional information is of benefit to prospective test takers. While I definitely hope more high school and undergraduate students are exposed to potential questions and concerns about prenatal testing and selective termination, I have concerns that addressing ethical, legal, and social issues of prenatal testing in health education or in the clinical encounter might be overwhelming in its sheer magnitude to prospective test takers, making arguably personal choices. As such, my work highlights the need for further research investigating how potential users want information, the types of information they want or think is missing, how to make materials comprehensive yet not overwhelming, and easiest to access. I think we will improve patient satisfaction and perhaps facilitate better outcomes if we provide patients in this context information that they believe they want along with information that we as professionals think that they should know.

For me, the public narrative about choice is one of my most interesting findings of my work. While I expected language about choice to be embedded in a broader public dialogue about abortion policy, I did not realize the complexity and nuance that would emerge publicly. Much attention focuses on personal choice. There is an implicit assumption that patients should be fully informed. However, this assumption does not place any parameters on how much information is sufficient for a patient to be fully informed or how presumably providers ought to go about fully informing patients (i.e., beyond legal documentation of informed consent). Another implicit assumption is that patients should freely exercise their choices. Unfortunately, this does not acknowledge real social and structural barriers to free choice. Ultimately, these assumption leave prospective parents with the responsibility of seeking information about prenatal testing and the ramifications of their choices. I find this problematic because I think it

overlooks potential fiduciary duties and social responsibilities. We cannot simply have personal choice without consideration for the duties and responsibilities.

With the diversity of choices expressed in the public narrative, I think we really need to explore which choices matter, in order to protect the rights that afford these choices. For example, choice often refers to patient options within healthcare decision-making, particularly an expansion of these options. Surely, there has to be limits with regards to these options. If we consider all pregnant woman should have access to a range of prenatal testing options, regardless of healthcare insurance coverage, costs and resources will decidedly limit our options. Alternatively, we might consider letting market forces dictate our use and nonuse. For another example, some might argue that more options means the availability of prenatal genetic testing for a wider array of conditions. Then, we might want to think more specifically about the types of genetic conditions for which we would want or consider prenatal testing. If we could in fact accurately and reliably test for adult onset genetic conditions, genetic susceptibilities or variants of uncertain significance, should we permit such testing in the prenatal context? I think these questions are important, and while not raised in the public narrative or specifically addressed in my work, I these questions deserve further consideration and attention, as well as further research.

While health messages highlight a consumer-driven healthcare marketplace, we need to have more transparent, public conversations about medical commons (Hiatt, 1975). If we take the popular frame that information is a good of genetic testing or more broadly of any medical testing, then is there a limit to the information that we might seek? There has to be. Yet, at least in the public discourse, this is largely absent. Frankly, we as individuals, providers, insurance companies, and policymakers make these decisions already. This is nothing new and happens in

nearly every facet of health, both in and out of the clinic. Consider a woman contemplating breast augmentation. If for personal or potentially social reasons, she wants this surgical intervention. She finds a provider, and the provider, based upon known insurance data and existing policies, would be able to tell her exactly how much it would cost her. Because this procedure is understood as elective, the patient would likely expect a substantial, personal, out-of-pocket cost. While an identical procedure might be performed on this same woman following a breast-cancer related lumpectomy or a burn injury at a different out-of-pocket cost, given likely different insurance reimbursement. I argue that this is the direction that genetics must venture and even strive towards. Information itself is not inherently good and is heavily value-laden. As such, we have to contextualize the information we might receive from genetic tests. Is the information so personally valuable that at least some patients would be willing to pay for the information out-of-pocket? If yes, then may be it is appropriate to do so, but charge them, rather than the medical commons. If a patient is not going to do anything with the information and not willing to pay for that information, is it really valuable? If we continue to use and emphasize a consumer-driven healthcare marketplace, then I think we have to let market forces drive this. However, I think that this approach demands greater transparency about costs.

If genetics, like other medical specialties, is to adopt this market-driven model, then I think we have to shift the current rhetoric and language. Based upon my analysis, health messages about genetic testing have co-opted language about early detection and disability prevention. Unlike surgery interventions, like the one mentioned above, the public understands this procedure is optional, unnecessary and motivated by personal desires; in fact, most in the public might even have difficulty identifying why breast augmentation might ever be necessary. From a communications perspective, this surgical language is genius: people who need surgery

can usually get reimbursed and the rest pay out-of-pocket costs. In contrast, the public health language implies a public good, suggesting that all people should have access. If we adopt this actually adopt this in practice, rather than just the language, the medical commons foots the bill for all genetic testing. If we accept that genetic tests are not equal, the meaning of the conditions for which we test evolves and that it is difficult to un-fund testing after previously funding it, then we should be particularly cautious about adopting a policy orientation that supports personal utility as a publicly valuable endpoint. To me, the most meaningful implications of my work here are: first, we need to change our rhetorical choices that conflate personal utility and traditional clinical utility or imply knowledge is inherently beneficial; second, we need to steer away from practice adoption that supports these underlying values, in the absence of tradition clinical utility. As a public health professional, I fully acknowledge the value of early detection and disability prevention; however, I also recognize the need for cost-effective strategies and approaches. Given more genetic knowledge and more genetic tests, I think that expanding traditional notions of clinical utility to include personal utility without understanding relevant contextual issues is at the very least unsustainable. More specifically, I would recommend that cell-free fetal DNA testing for Down syndrome remain an elective test with associate out-of-pocket costs. If we value this earlier, more precise screening, then I think we ought to be able to put a monetary cost on it. Given the availability of existing screening and valid, relatively low-risk diagnostic testing. I do not think that earlier, more precise screening is necessary. Such testing itself does not improve personal or population health outcomes. I hope that in 10 years the notion of elective genetic testing readily circulates in health messages.

At a policy level, I think there are some practical lessons to learn, when considering prenatal testing for Down syndrome as an exemplar for other genetic tests. First, genetic tests

are not equal. Each test, even each methodology, has different analytic validity, different purposes, and different desired endpoints; purposes and endpoint might even differ in the minds of individual patients and providers. Second, we have to consider the meaning of the conditions for which we can test may evolve with time. This may result from both medical interventions and social changes. In the 1970s, medical and popular views about living with Down syndrome were far more negative and severe than they are now. I would even argue that if we had just now discovered maternal serum testing, then the policies about screening might be different than they were when first introduced. Third, within our current healthcare infrastructure, it is difficult to restrict access via particular guidelines and policies once offered. For example, despite changing perspectives about prenatal testing for Down syndrome, there is no efforts -to the best of my knowledge- to restrict access or change policies to make such testing considered elective. For lack of a better analogy, it is hard to get the genie back into the bottle, even with evidence to do so. Because of this, I contend that we need to be particularly cautious as we translate research into practice. We need to ensure that we have sufficient evidence, reproducible by researchers without conflicts of interest and arguably even population-based studies, before making significant policy or practice changes. Using cell-free fetal DNA testing for Down syndrome as an example, we have seen that the early results offered much promise and initiated practice changes promoting adoption of such tests, for which we now have evidence of more false positive risk than originally predicted (Snyder *et al.*). This is not to say that the corporate involvement necessarily yield inaccurate data or allege misconduct, but rather to highlight that we know that there are differences between typical study populations and the general population. Individual and population health can be hurt in doing so. We also see that health messages about translational research garner much media attention, and in doing so, create a public narrative that

is also difficult to redirect. While I am not necessarily saying that we need to justify discretionary decisions using a precautionary principle, where changing the course of action requires proof of no harm, I do think that we have to be more cautious and more aware of the implications that such emerging technologies. Namely, I think that this means we need to ensure adequate funding of post-market studies about recently translated medical tests, including collection of information about psychosocial costs and financial ones.

RECOMMENDATIONS

Mass media can skew information about particular health care tests and procedures, particularly ones newly translating from bench-to-bedside. Even educated, information-seeking patients may have biased or incomplete knowledge about the testing options available to them. Health education and health communication materials outside the clinical encounter should be created to provide information about both risks and benefits of testing, as well as potential outcomes of specific prenatal testing methodologies. When contemplating pregnancy or at the latest during the first prenatal visit, clinicians should discuss patients' goals of care, in efforts to identify personal preferences and to support patient-centered care; this might take into consideration patient perspectives about utility (e.g., risk-benefit assessment or early detection/disability prevention) or values about choice (e.g., personal tendencies to want lots information and seek it, or concerns about too many choices or medicalization).

Prior to a clinical encounter where such tests might be offered, we should assess patient knowledge of potential risks and benefits. Patients should know the bottom line, out-of-pocket costs before clinical tests or procedures are administered, especially for recently translated technologies, which may not be fully covered by all health insurance. We should also help patients to consider their attitudes and values towards disability and pregnancy termination.

Seeking information about values is not about persuading patients in a particular way, but rather about ensuring patients have accurate, up-to-date, complete, unbiased information. In the public sphere, much misinformation and conflicting information exists, and at the very least we should strive to minimize confusion and anxiety. In instances where patients have inadequate information, we should offer information or resources to supplement pretest knowledge, so that patients are adequately understand risks and benefits and feel informed with the information they want to make their decisions. This does not necessarily mean creating new or personalized education materials, but rather informing patients of easily accessible, existing materials. In this manner, patients will be better able to assess whether a test is right for them in terms of their personal assessment.

References

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.

Hiatt, H. H. (1975). Protecting the medical commons: who is responsible? *The New England journal of medicine, 293*(5), 235-241. doi: 10.1056/nejm197507312930506

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Shendure, J. (2012). Noninvasive whole-genome sequencing of a human fetus. *Sci Transl Med, 4*(137), 137ra176. doi: 10.1126/scitranslmed.3004323

Snyder, M. W., Simmons, L. E., Kitzman, J. O., Coe, B. P., Henson, J. M., Daza, R. M., . . . Gammill, H. S. Copy-number variation and false positive prenatal aneuploidy screening results. *New England Journal of Medicine*.

Thomas, K., Bourke, J., Girdler, S., Bebbington, A., Jacoby, P., & Leonard, H. (2011). Variation Over Time in Medical Conditions and Health Service Utilization of Children with Down Syndrome. *The Journal of Pediatrics, 158*(2), 194-200.e191. doi: http://dx.doi.org/10.1016/j.jpeds.2010.08.045

BIBLIOGRAPHY

Chapter 1

Alderson, P. (2001). Down's syndrome: cost, quality and value of life. Social Science & Medicine (1982), 53(5), 627-638.

American College of Obstetricians and Gynecologists. (2007a). Screening for Fetal Chromosomal Abnormalities. Obstet Gynecol, 109(1).

American College of Obstetricians and Gynecologists. (2007b). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

American College of Obstetricians and Gynecologists. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm

American College of Obstetricians and Gynecologists. (2007d). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. Obstet Gynecol, 110(6), 1459.

American College of Obstetricians and Gynecologists. (2012). Noninvasive prenatal testing for fetal aneuploidy. Obstet Gynecol, 120(545), 1532–1534.

Americans with Disabilities Act, Pub. L. No. 110-325 (1990).

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. American Journal of Public Health, 89(11), 1649-1657.

Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.

Asch, A. (2003). Disability equality and prenatal testing: contradictory or compatible? Florida State University Law Review. Florida State University. College Of Law, 30(2), 315-342.

Baily, M. A., & Murray, T. H. (2008). Ethics, evidence, and cost in newborn screening. Hastings Center Report, 38(3), 23-31.

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? The American Journal of Bioethics, 9(8), 48-56.

Biesecker, B. B., & Hamby, L. (2000). What difference the disability community arguments should make for the delivery of prenatal genetic information. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.

Bindra, R., Heath, V., Liao, A., Spencer, K., & Nicolaides, K. H. (2002). One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. Ultrasound in Obstetrics and Gynecology, 20(3), 219-225. doi: 10.1046/j.1469-0705.2002.00808.x

Botkin, J. R., Clayton, E. W., Fost, N. C., Burke, W., Murray, T. H., Baily, M. A., . . . Ross, L. F. (2006). Newborn Screening Technology: Proceed With Caution. Pediatrics, 117(5), 1793-1799. doi: 10.1542/peds.2005-2547

Browner, C. H. (2000). Situating Women's Reproductive Activities. American Anthropologist, 102(4), 773-788.

Browner, C. H., & Press, N. (1996). The production of authoritative knowledge in American prenatal care. Medical Anthropology Quarterly, 10(2), 141-156.
Browner, C. H., & Press, N. A. (1995). The normalization of prenatal diagnostic screening. Conceiving the new world order: The global politics of reproduction, 307-322.
Buchanan, A., Brock, D. W., Daniels, N., & Wikler, D. (2000). From Chance to Choice: Genetics and Justice. Cambridge: Cambridge Univ Pr.
Burke, W., Atkins, D., Gwinn, M., Guttmacher, A., Haddow, J., Lau, J., . . . Wiesner, G. L. (2002). Genetic Test Evaluation: Information Needs of Clinicians, Policy Makers, and the Public. American Journal of Epidemiology, 156(4), 311-318. doi: 10.1093/aje/kwf055
Chao, S., Roberts, J. S., Marteau, T. M., Silliman, R., Cupples, L. A., & Green, R. C. (2008). Health behavior changes after genetic risk assessment for Alzheimer disease: The REVEAL Study. Alzheimer disease and associated disorders, 22(1).
Childress, J. F., Faden, R. R., Gaare, R. D., Gostin, L. O., Kahn, J., Bonnie, R. J., . . . Nieburg, P. (2002). Public health ethics: mapping the terrain. The Journal of Law, Medicine & Ethics, 30(2), 170-178.
Constantin, C. M., Faucett, A., & Lubin, I. M. (2005). A primer on genetic testing. Journal of Midwifery & Women's Health, 50(3), 197-204.
Cushing, P., & Smith, T. (2009). Multinational Review of English Language Disability Studies Degrees and Courses. Disability Studies Quarterly, 29(3).
Davis, D. S. (2010). Genetic dilemmas : reproductive technology, parental choices, and children's futures. Oxford; New York: Oxford University Press.
Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. American Journal of Obstetrics and Gynecology, 191(3), 1044-1048.
Gee, J. P. (2010). An introduction to discourse analysis : theory and method. New York [etc.]: Routledge.
Ginsburg, F., & Rapp, R. (1999). Fetal reflections: Confessions of two feminist anthropologists as mutual informats. In L. M. Morgan & M. W. Michaels (Eds.), Fetal subjects, feminist positions. Philadelphia: University of Pennsylvania Press.
Green, R. C., Roberts, J. S., Cupples, L. A., Relkin, N. R., Whitehouse, P. J., Brown, T., . . . Group, R. S. (2009). Disclosure of APOE genotype for risk of Alzheimer's disease. The New England journal of medicine, 361(3), 245-254.
Groce, N. E. (1985). Everyone here spoke sign language : hereditary deafness on Martha's Vineyard. Cambridge, Mass.: Harvard University Press.
Haller, B. A. (2010). Representing disability in an ableist world : essays on mass media. Louisville, KY: The Advocado Press.
Ivry, T. (2008). At the Back Stage of Prenatal Care: Japanese Ob-Gyns Negotiating Prenatal Diagnosis. Medical Anthropology Quarterly, 20(4), 441-468.
Kalfoglou, A., Suthers, K., Scott, J., & Hudson, K. (2004). Reproductive genetic testing: What America thinks. Washington, DC: Genetics and Public Policy Center.
Kelly, S., & Farrimond, H. (2012). Non-invasive prenatal genetic testing: a study of public attitudes. Public Health Genomics, 15(2), 73-81.
Khoury, M., McBride, C., Schully, S., Ioannidis, J. P., Feero, W. G., Janssens, A. C. J., . . . Cargill, M. (2009). The scientific foundation for personal genomics: recommendations from a National Institutes of Health–Centers for Disease Control and Prevention multidisciplinary workshop. Genetics in Medicine, 11(8), 559.

Khoury, M. J., Jones, K., & Grosse, S. D. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. Genetics in Medicine, 8(3), 191.
Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: conversations with my son. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.
Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. Science translational medicine, 4(137), 137ra176-137ra176.
Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". AJOB Primary Research, 2(2), 8-20.
Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing. Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70(1), 7-9.
LaRusse, S., Roberts, J. S., Marteau, T. M., Katzen, H., Linnenbringer, E. L., Barber, M., . . . Relkin, N. R. (2005). Genetic susceptibility testing versus family history-based risk assessment: Impact on perceived risk of Alzheimer disease. Genetics in Medicine, 7(1), 48-53.
LeGreco, M., & Tracy, S. J. (2009). Discourse tracing as qualitative practice. Qualitative Inquiry, 15(9), 1516-1543.
Leung, T. Y., Chan, L. W., Law, L. W., Sahota, D. S., Fung, T. Y., Leung, T. N., & Lau, T. K. (2009). First trimester combined screening for Trisomy 21 in Hong Kong: outcome of the first 10,000 cases. The Journal of Maternal-Fetal & Neonatal Medicine, 22(4), 300-304. doi: 10.1080/14767050802430842
Lippman, A. (1999). Embodied knowledge and making sense of prenatal diagnosis. Journal of genetic counseling, 8(5), 255-274.
Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. American Journal of Human Genetics, 51(4), 936.
Lo, Y., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W., & Wainscoat, J. S. (1997). Presence of fetal DNA in maternal plasma and serum. The Lancet, 350(9076), 485-487.
Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. The American Journal of Human Genetics, 62(4), 768-775.
Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: a commentary. American journal of medical genetics. Part A, 155A(8), 1777-1785.
Malone, F. D., Canick, J. A., Ball, R. H., Nyberg, D. A., Comstock, C. H., Bukowski, R., . . . D'Alton, M. E. (2005). First-Trimester or Second-Trimester Screening, or Both, for Down's Syndrome. New England Journal of Medicine, 353(19), 2001-2011. doi: doi:10.1056/NEJMoa043693
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. Prenatal diagnosis, 19(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Markel, H. (1997). Scientific advances and social risks: historical perspectives of genetic screening programs for Sickle Cell Disease, Tay-Sachs Disease, neural tube defects and Down Syndrome, 1970-1997. Medical and Public Health Law. http://biotech.law.lsu.edu/research/fed/tfgt/appendix6.htm

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. Sociology of health & illness, 32(1), 37-56.

Markens, S., Browner, C. H., & Press, N. A. (1999). Because of the risks': how US pregnant women account for refusing prenatal screening. Social Science & Medicine, 49(3), 359-369.

Marteau, T., Drake, H., & Bobrow, M. (1994). Counselling following diagnosis of a fetal abnormality: the differing approaches of obstetricians, clinical geneticists, and genetic nurses. Journal of medical genetics, 31(11), 864-867.

Marteau, T., Plenicar, M., & Kidd, J. (1993). Obstetricians presenting amniocentesis to pregnant women: practice observed. Journal of Reproductive and Infant Psychology, 11(1), 3-10.

Miron-Shatz, T., Hanoch, Y., Graef, D., & Sagi, M. (2009). Presentation Format Affects Comprehension and Risk Assessment: The Case of Prenatal Screening. Journal of Health Communication, 14(5), 439-450. doi: 10.1080/10810730903032986

Murkoff, H. E., & Mazel, S. (2008). What to expect when you're expecting (4th ed.). New York: Workman Pub.

Nadler, H. L. (1968). Antenatal detection of hereditary disorders. Pediatrics, 42(6), 912-918.

National Coalition for Health Professional Education in Genetics, & National Society of Genetic Counselors. (2012). Prenatal Screening/Testing Options for Trisomy 21 and Other Fetal Aneuploidies. In N.-I. P. T. N. Factsheet (Ed.).

Nelson, J. L. (1998). The meaning of the act: reflections on the expressive force of reproductive decision making and policies. Kennedy Inst Ethics J, 8(2), 165-182.

Nicolaides, K. H. (2011). Screening for fetal aneuploidies at 11 to 13 weeks. Prenatal diagnosis, 31(1), 7-15. doi: 10.1002/pd.2637

Nicolaides, K. H., Spencer, K., Avgidou, K., Faiola, S., & Falcon, O. (2005). Multicenter study of first-trimester screening for trisomy 21 in 75 821 pregnancies: results and estimation of the potential impact of individual risk-orientated two-stage first-trimester screening. Ultrasound in Obstetrics and Gynecology, 25(3), 221-226. doi: 10.1002/uog.1860

Norton, M. E., Brar, H., Weiss, J., Karimi, A., Laurent, L. C., Caughey, A. B., . . . Song, K. (2012). Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18. American Journal of Obstetrics and Gynecology, 207(2), 137.e131-137.e138. doi: http://dx.doi.org/10.1016/j.ajog.2012.05.021

Oliver, M. (1990). The individual and social models of disability. Paper presented at the Joint Workshop of the Living Options Group and the Research Unit of the Royal College of Physicians.

Oliver, M. (1996). Defining Impairment and Disability: Issues at Stake. Paper presented at the Exploring the Divide, University of Leeds.

Ormond, K. E., Gill, C. J., Semik, P., & Kirschner, K. L. (2003). Attitudes of health care trainees about genetics and disability: Issues of access, health care communication, and decision making. Journal of genetic counseling, 12(4), 333-349.

Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genetics in Medicine, 13(11), 913-920.

Pilnick, A. (2004). 'It's Just One of the Best Tests that We've Got at the Moment': the Presentation of Nuchal Translucency Screening for Fetal Abnormality in Pregnancy. Discourse & Society, 15(4), 451-465. doi: 10.1177/0957926504043710

Press, N., & Browner, C. (1994). Collective silences, collective fictions. Women and Prenatal Testing: Facing the Challenges of Genetic Technology, 209-210.

Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. Social Science and Medicine, 45(7), 979-989.

Press, N., Wilfond, B., Murray, M., & Burke, W. (2011). The power of knowledge: How carrier and prenatal screening altered the clinical goals of genetic testing. In W. Burke, K. A. Edwards, S. Goering, S. Holland & S. B. Trinidad (Eds.), Achieving Justice in Genomic Translation: Re-Thinking the Pathway to Benefit: Re-Thinking the Pathway to Benefit: Oxford University Press, USA.

Rapp, R. (1993). Amniocentesis in sociocultural perspective. Journal of genetic counseling, 2(3), 183-196.

Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), Representations of motherhood: Yale University Press.

Rapp, R. (1998). Refusing prenatal diagnosis: The meanings of bioscience in a multicultural world. Science, technology & human values, 23(1), 45-70.

Rapp, R. (1999). Testing women, testing the fetus: The social impact of amniocentesis in America.

Riley, C. A. (2005). Disability and the media: prescriptions for change. Hanover, NH: University Press of New England.

Roberts, J. S., Barber, M., Brown, T. M., Cupples, L. A., Farrer, L. A., LaRusse, S. A., . . . Green, R. C. (2004). Who seeks genetic susceptibility testing for Alzheimer's disease? Findings from a multisite, randomized clinical trial. Genetics in Medicine, 6(4).

Rothman, B. K. (1994). The Tentative Pregnancy: Then and Now. In K. H. Rothenberg & E. J. Thomson (Eds.), Women and prenatal testing : facing the challenges of genetic technology. Columbus: Ohio State University Press.

Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), The disability studies reader (2nd ed., pp. xviii, 451 p.). New York: Routledge.

Secretary's Advisory Committee on Genetic Testing. (2000). Enhancing the oversight of genetic tests: recommendations of the SACGT. from http://oba.od.nih.gov/oba/sacgt/highlights/sacgt012700summ.pdf

Shapiro, J. P. (1993). No pity : people with disabilities forging a new civil rights movement. New York: Times Books.

Skotko, B. G. (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? Archives of disease in childhood, 94(11), 823-826.

Spencer, K., Souter, V., Tul, N., Snijders, R., & Nicolaides, K. H. (1999). A screening program for trisomy 21 at 10–14 weeks using fetal nuchal translucency, maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A. Ultrasound in Obstetrics and Gynecology, 13(4), 231-237. doi: 10.1046/j.1469-0705.1999.13040231.x

Steinbock, B. (2000). Disability, Prenatal Testing, and Selective Abortion. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.
Stern, A. M. (2005). Sterilized in the name of public health: race, immigration, and reproductive control in modern California. American Journal of Public Health, 95(7), 1128.
Task Force on Genetic Testing, Holtzman, N. A., & Watson, M. S. (1998). Promoting safe and effective genetic testing in the United States : final report of the Task Force on Genetic Testing. Baltimore: Johns Hopkins University Press.
Taylor, J. S. (2000). Of sonograms and baby prams: Prenatal diagnosis, pregnancy, and consumption. Feminist Studies, 26(2), 391-418.
Ten Great Public Health Achievements--United States, 1900-1999. (1999). JAMA: The Journal of the American Medical Association, 281(16), 1481-1481. doi: 10.1001/jama.281.16.1481
US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml
Vanzwieten, M., Willems, D., Knegt, L., & Leschot, N. (2006). Communication with patients during the prenatal testing procedure: An explorative qualitative study. Patient Educ Couns, 63(1-2), 161-168. doi: 10.1016/j.pec.2005.09.013
Wald, N. J., Huttly, W. J., & Hackshaw, A. K. (2003). Antenatal screening for Down's syndrome with the quadruple test. The Lancet, 361(9360), 835-836. doi: http://dx.doi.org/10.1016/S0140-6736(03)12680-3
Wald, N. J., Huttly, W. J., Murphy, K. W., K, A., Bestwick, J. P., & Rodeck, C. H. (2009). Antenatal screening for Down's syndrome using the Integrated test at two London hospitals. Journal of Medical Screening, 16(1), 7-10. doi: 10.1258/jms.2009.008094
Zick, C. D., Mathews, C. J., Roberts, J. S., Cook-Deegan, R., Pokorski, R. J., & Green, R. C. (2005). Genetic Testing For Alzheimer's Disease And Its Impact On Insurance Purchasing Behavior. Health Affairs, 24(2), 483-490. doi: 10.1377/hlthaff.24.2.483
Zimmern, R. L., & Kroese, M. (2007). The evaluation of genetic tests. Journal of Public Health, 29(3), 246-250. doi: 10.1093/pubmed/fdm028

Chapter 2

ACOG Office of Communications. (2007a). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm
ACOG Office of Communications. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm
Aramesh, K. (2009). A closer look at the abortion debate in Iran. The American Journal of Bioethics, 9(8), 57-58.
Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. American Journal of Public Health, 89(11), 1649-1657.
Asch, A., & Wasserman, D. (2005). Where Is the Sin in Synecdoche? Prenatal Testing and the Parent-Child Relationship. In D. T. Wasserman, R. S. Wachbroit & J. E. Bickenbach (Eds.), Quality of life and human difference : genetic testing, health care, and disability. New York: Cambridge University Press.
Azzolino v. Dingfelder, No. No. 718PA84, 337 528 (NC: Supreme Court 1985).

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? The American Journal of Bioethics, 9(8), 48-56.
Beauchamp, T. L., & Childress, J. F. (1994). Principles of biomedical ethics. New York: Oxford University Press.
Bérubé, M. (1998). Life as we know it : a father, a family, and an exceptional child. New York: Vintage Books.
Buccafurni, D., & Chang, P. L. (2009). Does prenatal diagnosis morally require provision of selective abortion? The American Journal of Bioethics, 9(8), 65-67.
Condit, C. M. (1990). Decoding abortion rhetoric : communicating social change. Urbana: University of Illinois Press.
Davis, D. S. (2010). Genetic dilemmas : reproductive technology, parental choices, and children's futures. Oxford; New York: Oxford University Press.
Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2012). Noninvasive prenatal testing/noninvasive prenatal diagnosis: The position of the national society of genetic counselors. Journal of genetic counseling, 1-5.
Disabled People's International. (2000). Disabled Persons Speak on the New Genetics.
Disabled People's International. (2012). from http://www.dpi.org/
Eddleman, K. A., Malone, F. D., Sullivan, L., Dukes, K., Berkowitz, R. L., Kharbutli, Y., . . . D'Alton, M. E. (2006). Pregnancy loss rates after midtrimester amniocentesis. Obstetrics and gynecology, 108(5), 1067-1072.
Gallagher v. Duke University, No. No. 87-1056, 852 773 (Court of Appeals, 4th Circuit 1988).
Gee, J. P. (2010). An introduction to discourse analysis : theory and method. New York [etc.]: Routledge.
Ginsburg, F., & Rapp, R. (2010). Enabling Disability: Rewriting Kinship, Reimagining Citizenship. In L. J. Davis (Ed.), The disability studies reader (3rd ed., pp. xviii, 451 p.). New York: Routledge.
Hahn, S., & Holzgreve, W. (2002). Prenatal diagnosis using fetal cells and cell-free fetal DNA in maternal blood: what is currently feasible? Clinical Obstetrics and Gynecology, 45(3), 649.
Harbeson v. Parke-Davis, Inc, No. No. 48331-1, 656 483 (Wash: Supreme Court 1983).
Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. Qual Health Res, 15(9), 1277-1288. doi: 10.1177/1049732305276687
Kon, A. A. (2009). Prenatal testing can be advantageous even when abortion is not an option. The American Journal of Bioethics, 9(8), 69-70.
Langlois, S., Brock, J.-A., & Genetics Committee. (2013). Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. J Obstet Gynaecol Can, 35(2), 177-181.
Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. American Journal of Human Genetics, 51(4), 936.
Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. The American Journal of Human Genetics, 62(4), 768-775.
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. Prenatal diagnosis, 19(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Morabia, A., & Zhang, F. (2004). History of medical screening: from concepts to action. Postgraduate medical journal, 80(946), 463-469.
Munro v. Regents of University of California, No. Docket No. B037779, 215 977 (Cal: Court of Appeal, 2nd Appellate Dist., 7th Div. 1989).
Nadler, H. L. (1968). Antenatal detection of hereditary disorders. Pediatrics, 42(6), 912-918.
Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. Health Expectations, 11(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x
Press, N., & Browner, C. (1994). Collective silences, collective fictions. Women and Prenatal Testing: Facing the Challenges of Genetic Technology, 209-210.
Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. Social Science and Medicine, 45(7), 979-989.
Procanik v. Cillo, 502 94 (NJ: Superior Court, Law Div. 1985).
Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), Representations of motherhood: Yale University Press.
Roe v. Wade, 410 113 (Supreme Court 1973).
Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), The disability studies reader (2nd ed., pp. xviii, 451 p.). New York: Routledge.
Seavilleklein, V. (2009). Challenging the rhetoric of choice in prenatal screening. Bioethics, 23(1), 68-77.
Smith, L. (1995). Antiabortion group's leaflets liken doctors to war criminals. Washington Post, p. 3.
Sperling, D. (2009). From Iran to Latin America: must prenatal diagnosis necessarily be provided with abortion for congenital abnormalities? The American Journal of Bioethics, 9(8), 61-63.
The Association of Faculties of Medicine of Canada Public Health Educators' Network. Basic concepts in prevention, surveillance and health promotion. In D. Donovan, B. McDowell & D. Hunter (Eds.), Primer on Population Health: The Association of the Faculties of Medicine of Canada. Retrieved from http://phprimer.afmc.ca/Part1-TheoryThinkingAboutHealth/Chapter4BasicConceptsInPreventionSurveillanceAndHealthPromotion/Thestagesofprevention.
The Noninvasive Prenatal Screening Work Group. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. Genetics in Medicine, 15(5), 395-398.
Turpin v. Sortini, No. Docket No. SF 24319, 643 954 (Cal: Supreme Court 1982).
US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml
Varvasovszky, Z., & Brugha, R. (2000). How to do (or not to do) . . . A stakeholder analysis. Health policy and planning, 15(3), 338-345.
Wasserman, D., & Asch, A. (2009). An unjustified exception to an unjust law? The American Journal of Bioethics, 9(8), 63-65.
Wilkinson, S. (2008). "Eugenics talk" and the language of bioethics. Journal of Medical Ethics, 34(6), 467-471. doi: 10.1136/jme.2007.021592
Zivotofsky, A. Z., & Jotkowitz, A. (2009). Prenatal diagnosis and abortion are not in conflict in Israel. The American Journal of Bioethics, 9(8), 58-60.

Chapter 3

Bakke, A. (2013). Disabling the Rhetoric of Prenatal Genetic Testing: A Critical Analysis of Online Self-Help Literature. Critical Disability Discourse, 5, 11-38.

Fox, S. (2012). Pew Internet: Health. Retrieved 24 January, 2013, from http://www.pewinternet.org/Commentary/2011/November/Pew-Internet-Health.aspx

Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. Qual Health Res, 15(9), 1277-1288. doi: 10.1177/1049732305276687

Kellogg, G., Slattery, L., Hudgins, L., & Ormond, K. (2014). Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing. Journal of genetic counseling, 23(5), 805-813. doi: 10.1007/s10897-014-9694-7

Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial Barriers on Use of Prenatal Genetic Testing. Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70(1), 7-9.

Lawson, K. L., Carlson, K., & Shynkaruk, J. M. (2012). The Portrayal of Down Syndrome in Prenatal Screening Information Pamphlets. J Obstet Gynaecol Can, 34(8), 760-768.

Lewis, C., Silcock, C., & Chitty, L. S. (2013). Non-invasive prenatal testing for Down's syndrome: pregnant women's views and likely uptake. Public Health Genomics, 16(5), 223-232. doi: 10.1159/000353523

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. Sociology of health & illness, 32(1), 37-56.

Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf

Moorhead, S. A., Hazlett, D. E., Harrison, L., Carroll, J. K., Irwin, A., & Hoving, C. (2013). A New Dimension of Health Care: Systematic Review of the Uses, Benefits, and Limitations of Social Media for Health Communication. Journal of Medical Internet Research, 15(4), e85. doi: 10.2196/jmir.1933

Muller, C., & Cameron, L. D. (2015). It's complicated – Factors predicting decisional conflict in prenatal diagnostic testing. Health Expectations, n/a-n/a. doi: 10.1111/hex.12363

Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. Health Expectations, 11(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x

Press, N., & Browner, C. (1994). Collective silences, collective fictions. Women and Prenatal Testing: Facing the Challenges of Genetic Technology, 209-210.

Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. Social Science and Medicine, 45(7), 979-989.

Reed, K. (2009). 'It's them faulty genes again': women, men and the gendered nature of genetic responsibility in prenatal blood screening. Sociology of health & illness, 31(3), 343-359. doi: 10.1111/j.1467-9566.2008.01134.x

Stapleton, H., Kirkham, M., & Thomas, G. (2002). Qualitative study of evidence based leaflets in maternity care. BMJ : British Medical Journal, 324(7338), 639-639.
van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. Patient Educ Couns, 63(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007

Chapter 4

ACOG Office of Communications. (2007). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. *ACOG News Release.* Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm
Amundson, R. (2005). Disability, Ideology, and Quality of Life: A Bias in Biomedical Ethics. In D. T. Wasserman, R. S. Wachbroit & J. E. Bickenbach (Eds.), *Quality of life and human difference : genetic testing, health care, and disability* (pp. 101-124). New York: Cambridge University Press.
Amundson, R., & Tresky, S. (2007). On a Bioethical Challenge to Disability Rights. *Journal of Medicine and Philosophy, 32*(6), 541-561. doi: 10.1080/03605310701680924
Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. *American Journal of Public Health, 89*(11), 1649-1657.
Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. *Prenatal testing and disability rights, 51*.
Asch, A., & Wasserman, D. (2005). Where is the sin in synecdoche. *Quality of life and human difference: Genetic testing, health care, and disability*.
Baglieri, S., & Shapiro, A. (2012). *Disability studies and the inclusive classroom: Critical practices for creating least restrictive attitudes*: Routledge.
Bubela, T. M., & Caulfield, T. A. (2004). Do the print media "hype" genetic research? A comparison of newspaper stories and peer-reviewed research papers. *Canadian Medical Association Journal, 170*(9), 1399-1407. doi: 10.1503/cmaj.1030762
Caulfield, T. (2004). Biotechnology and the popular press: hype and the selling of science. *TRENDS in biotechnology, 22*(7), 337-339.
Davis, D. S. (2010). Genetic Dilemmas: Reproductive Technology, Parental Choices, and Children's Futures.
de Jong, A., Dondorp, W. J., Frints, S. G., de Die-Smulders, C. E., & de Wert, G. M. (2011). Advances in prenatal screening: the ethical dimension. *Nature Reviews Genetics, 12*(9), 657-663.
Devaney, S. A., Palomaki, G. E., Scott, J. A., & Bianchi, D. (2011). Noninvasive fetal sex determination using cell-free fetal DNA: a systematic review and meta-analysis. *JAMA, 306*(6), 627-636.
Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2013). Noninvasive prenatal testing/noninvasive prenatal diagnosis: the position of the National Society of Genetic Counselors. *Journal of genetic counseling, 22*(3), 291-295.
Dolan, G., Iredale, R., Williams, R., & Ameen, J. (2004). Consumer use of the internet for health information: a survey of primary care patients. *International Journal of Consumer Studies, 28*(2), 147-153.
Etchegary, H., Potter, B., Howley, H., Cappelli, M., Coyle, D., Graham, I., . . . Wilson, B. (2008). The influence of experiential knowledge on prenatal screening and testing decisions. *Genetic testing, 12*(1), 115-124.

Garland-Thomson, R. (2005). Disability and Representation. *PMLA, 120*(2), 522-527.
Gigerenzer, G., Gaissmaier, W., Kurz-Milcke, E., Schwartz, L. M., & Woloshin, S. (2007). Helping doctors and patients make sense of health statistics. *Psychological science in the public interest, 8*(2), 53-96.
Haller, B. A. (2010). *Representing disability in an ableist world : essays on mass media*. Louisville, KY: The Advocado Press.
Holtzman, N. A., Bernhardt, B. A., Mountcastle-Shah, E., Rodgers, J. E., Tambor, E., & Geller, G. (2005). The quality of media reports on discoveries related to human genetic diseases. *Public Health Genomics, 8*(3), 133-144.
Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. *Qual Health Res, 15*(9), 1277-1288. doi: 10.1177/1049732305276687
Hubbard, R. (2010). Abortion and disability: who should and who should not inhabit the world. In L. J. Davis (Ed.), *The Disability Studies Reader* (3 ed., pp. 107-119). New York, NY: Routledge.
Inhorn, M. C. (2006). Defining women's health: A dozen messages from more than 150 ethnographies. *Medical Anthropology Quarterly, 20*(3), 345-378.
Ivry, T. (2009). *Embodying culture: pregnancy in Japan and Israel*: Rutgers University Press.
Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: Conversations with my son. In E. Parens & A. Asch (Eds.), *Prenatal testing and disability rights* (pp. 165-195): Georgetown UP.
Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. *Science translational medicine, 4*(137), 137ra176-137ra176.
Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". *AJOB Primary Research, 2*(2), 8-20.
Langlois, S., Brock, J.-A., Wilson, R. D., Audibert, F., Carroll, J., Cartier, L., . . . Senikas, V. (2013). Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. *J Obstet Gynaecol Can, 35*(2), 177-181.
Lindemann, H., & Nelson, J. L. (2008). The romance of the family. *Hastings Center Report, 38*(4), 19-21.
Linton, S. (1998). *Claiming disability : knowledge and identity*. New York: New York University Press.
Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. *American journal of human genetics, 51*(4), 936.
Lo, Y., Tein, M. S., Lau, T. K., Haines, C. J., Leung, T. N., Poon, P. M., . . . Hjelm, N. M. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. *The American Journal of Human Genetics, 62*(4), 768-775.
Lulofs, N. (2013). Top 25 U.S. Newspapers for March 2013. Retrieved 08-27, 2014, from http://www.auditedmedia.com/news/blog/top-25-us-newspapers-for-march-2013.aspx
Lyerly, A. D., Mitchell, L. M., Armstrong, E. M., Harris, L. H., Kukla, R., Kuppermann, M., & Little, M. O. (2007). Risks, values, and decision making surrounding pregnancy. *Obstetrics & Gynecology, 109*(4), 979-984.
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a

systematic literature review. *Prenatal diagnosis, 19*(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. *Sociology of health & illness, 32*(1), 37-56.

McCoyd, J. L. (2010). Authoritative knowledge, the technological imperative and women's responses to prenatal diagnostic technologies. *Culture, Medicine, and Psychiatry, 34*(4), 590-614.

Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf

Mills, C. B., & Erzikova, E. (2012). Prenatal Testing, Disability, and Termination: An Examination of Newspaper Framing. *Disability Studies Quarterly, 32*(3).

Moynihan, R., Bero, L., Ross-Degnan, D., Henry, D., Lee, K., Watkins, J., . . . Soumerai, S. B. (2000). Coverage by the news media of the benefits and risks of medications. *New England Journal of Medicine, 342*(22), 1645-1650.

Moynihan, R., Heath, I., & Henry, D. (2002). Selling sickness: the pharmaceutical industry and disease mongeringCommentary: Medicalisation of risk factors. *Bmj, 324*(7342), 886-891.

Natoli, J. L., Ackerman, D. L., McDermott, S., & Edwards, J. G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). *Prenatal diagnosis, 32*(2), 142-153.

Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine, 13*(11), 913-920.

Pew Research Center. (2010a). Americans Spending More Time Following the News. Retrieved 08-26, 2014, from http://www.people-press.org/files/legacy-pdf/652.pdf

Pew Research Center. (2010c). How News Happens. Retrieved 08-26, 2014, from http://www.journalism.org/2010/01/11/how-news-happens/

Pew Research Center. (2012). Where Americans Get News. Retrieved 08-26, 2014, from http://www.journalism.org/media-indicators/where-americans-get-news/

Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Boland, I. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. *Health Expectations, 11*(4), 355-365.

President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. (1983). *Screening and Counseling for Genetic Conditions: A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs*. Washington, DC: U. S. Government Printing Office Retrieved from http://bioethics.georgetown.edu/pcbe/reports/past_commissions/geneticscreening.pdf.

Press, N. A. (2008). Genetic Testing and Screening. *From Birth to Death and Bench to Clinic: The Hastings Center Bioethics Briefing Book for Journalists, Policymakers, and Campaigns*, 73-78.

Press, N. A., & Browner, C. H. (1994). Collective silences, collective fictions. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*, 209-210.
Press, N. A., & Browner, C. H. (1997a). Why Women Say Yes to Prenatal Diagnosis. *Social Science and Medicine, 45*(7), 979-989.
Press, N. A., & Browner, C. H. (1997c). Why women say yes to prenatal diagnosis. *Social Science & Medicine, 45*(7), 979-989.
Rapp, R. (1999). *Testing women, testing the fetus: The social impact of amniocentesis in America*.
Riley, C. A. (2005). *Disability and the media: prescriptions for change*. Hanover, NH: University Press of New England.
Saxton, M. (2010). Disability rights and selective abortion. In L. J. Davis (Ed.), *The Disability Studies Reader* (3 ed., pp. 120-132). New York, NY: Routledge.
Schwitzer, G. (2008). How do US journalists cover treatments, tests, products, and procedures? An evaluation of 500 stories. *PLoS medicine, 5*(5), e95.
Scully, J. L. (2008). Disability and genetics in the era of genomic medicine. *Nature Reviews Genetics, 9*(10), 797-802.
Seavilleklein, V. (2009). Challenging the rhetoric of choice in prenatal screening. *Bioethics, 23*(1), 68-77.
Shapiro, J. P. (1993). *No pity : people with disabilities forging a new civil rights movement*. New York: Times Books.
Shuchman, M., & Wilkes, M. S. (1997). Medical scientists and health news reporting: a case of miscommunication. *Ann Intern Med, 126*(12), 976-982.
St-Jacques, S., Grenier, S., Charland, M., Forest, J. C., Rousseau, F., & Légaré, F. (2008). Decisional needs assessment regarding Down syndrome prenatal testing: a systematic review of the perceptions of women, their partners and health professionals. *Prenatal diagnosis, 28*(13), 1183-1203.
Taylor, A. (2013). " Lives Worth Living:" Theorizing Moral Status and Expressions of Human Life. *Disability Studies Quarterly, 33*(4).
The Noninvasive Prenatal Screening Work Group. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. *Genetics in Medicine, 15*(5), 395-398.
Tremain, S. (2006). Reproductive Freedom, Self-Regulation, and the Government of Impairment in Utero. *Hypatia, 21*(1), 35-53. doi: 10.2307/3811076
van den Berg, M., Timmermans, D. R., Ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2005). Are pregnant women making informed choices about prenatal screening? *Genet Med, 7*(5), 332-338.
van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. *Patient Educ Couns, 63*(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007
van den Heuvel, A., Chitty, L., Dormandy, E., Newson, A., Deans, Z., Attwood, S., . . . Marteau, T. M. (2010). Will the introduction of non-invasive prenatal diagnostic testing erode informed choices? An experimental study of health care professionals. *Patient Educ Couns, 78*(1), 24-28. doi: 10.1016/j.pec.2009.05.014
Wilson, A., Bonevski, B., Jones, A., & Henry, D. (2009). Media reporting of health interventions: signs of improvement, but major problems persist. *PLoS One, 4*(3), ACOG Office of Communications. (2007). ACOG's Screening Guidelines on Chromosomal

Abnormalities What They Mean to Patients and Physicians. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm
Amundson, R. (2005). Disability, Ideology, and Quality of Life: A Bias in Biomedical Ethics. In D. T. Wasserman, R. S. Wachbroit & J. E. Bickenbach (Eds.), Quality of life and human difference : genetic testing, health care, and disability (pp. 101-124). New York: Cambridge University Press.
Amundson, R., & Tresky, S. (2007). On a Bioethical Challenge to Disability Rights. Journal of Medicine and Philosophy, 32(6), 541-561. doi: 10.1080/03605310701680924
Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. American Journal of Public Health, 89(11), 1649-1657.
Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. Prenatal testing and disability rights, 51.
Asch, A., & Wasserman, D. (2005). Where is the sin in synecdoche. Quality of life and human difference: Genetic testing, health care, and disability.
Baglieri, S., & Shapiro, A. (2012). Disability studies and the inclusive classroom: Critical practices for creating least restrictive attitudes: Routledge.
Bubela, T. M., & Caulfield, T. A. (2004). Do the print media "hype" genetic research? A comparison of newspaper stories and peer-reviewed research papers. Canadian Medical Association Journal, 170(9), 1399-1407. doi: 10.1503/cmaj.1030762
Caulfield, T. (2004). Biotechnology and the popular press: hype and the selling of science. TRENDS in biotechnology, 22(7), 337-339.
Davis, D. S. (2010). Genetic Dilemmas: Reproductive Technology, Parental Choices, and Children's Futures.
de Jong, A., Dondorp, W. J., Frints, S. G., de Die-Smulders, C. E., & de Wert, G. M. (2011). Advances in prenatal screening: the ethical dimension. Nature Reviews Genetics, 12(9), 657-663.
Devaney, S. A., Palomaki, G. E., Scott, J. A., & Bianchi, D. (2011). Noninvasive fetal sex determination using cell-free fetal DNA: a systematic review and meta-analysis. JAMA, 306(6), 627-636.
Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2013). Noninvasive prenatal testing/noninvasive prenatal diagnosis: the position of the National Society of Genetic Counselors. Journal of genetic counseling, 22(3), 291-295.
Dolan, G., Iredale, R., Williams, R., & Ameen, J. (2004). Consumer use of the internet for health information: a survey of primary care patients. International Journal of Consumer Studies, 28(2), 147-153.
Etchegary, H., Potter, B., Howley, H., Cappelli, M., Coyle, D., Graham, I., . . . Wilson, B. (2008). The influence of experiential knowledge on prenatal screening and testing decisions. Genetic testing, 12(1), 115-124.
Garland-Thomson, R. (2005). Disability and Representation. PMLA, 120(2), 522-527.
Gigerenzer, G., Gaissmaier, W., Kurz-Milcke, E., Schwartz, L. M., & Woloshin, S. (2007). Helping doctors and patients make sense of health statistics. Psychological science in the public interest, 8(2), 53-96.
Haller, B. A. (2010). Representing disability in an ableist world : essays on mass media. Louisville, KY: The Advocado Press.

Holtzman, N. A., Bernhardt, B. A., Mountcastle-Shah, E., Rodgers, J. E., Tambor, E., & Geller, G. (2005). The quality of media reports on discoveries related to human genetic diseases. Public Health Genomics, 8(3), 133-144.

Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. Qual Health Res, 15(9), 1277-1288. doi: 10.1177/1049732305276687

Hubbard, R. (2010). Abortion and disability: who should and who should not inhabit the world. In L. J. Davis (Ed.), The Disability Studies Reader (3 ed., pp. 107-119). New York, NY: Routledge.

Inhorn, M. C. (2006). Defining women's health: A dozen messages from more than 150 ethnographies. Medical Anthropology Quarterly, 20(3), 345-378.

Ivry, T. (2009). Embodying culture: pregnancy in Japan and Israel: Rutgers University Press.

Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: Conversations with my son. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights (pp. 165-195): Georgetown UP.

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. Science translational medicine, 4(137), 137ra176-137ra176.

Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". AJOB Primary Research, 2(2), 8-20.

Langlois, S., Brock, J.-A., Wilson, R. D., Audibert, F., Carroll, J., Cartier, L., . . . Senikas, V. (2013). Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. J Obstet Gynaecol Can, 35(2), 177-181.

Lindemann, H., & Nelson, J. L. (2008). The romance of the family. Hastings Center Report, 38(4), 19-21.

Linton, S. (1998). Claiming disability : knowledge and identity. New York: New York University Press.

Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. American journal of human genetics, 51(4), 936.

Lo, Y., Tein, M. S., Lau, T. K., Haines, C. J., Leung, T. N., Poon, P. M., . . . Hjelm, N. M. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. The American Journal of Human Genetics, 62(4), 768-775.

Lulofs, N. (2013). Top 25 U.S. Newspapers for March 2013. Retrieved 08-27, 2014, from http://www.auditedmedia.com/news/blog/top-25-us-newspapers-for-march-2013.aspx

Lyerly, A. D., Mitchell, L. M., Armstrong, E. M., Harris, L. H., Kukla, R., Kuppermann, M., & Little, M. O. (2007). Risks, values, and decision making surrounding pregnancy. Obstetrics & Gynecology, 109(4), 979-984.

Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. Prenatal diagnosis, 19(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b

Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. Sociology of health & illness, 32(1), 37-56.

McCoyd, J. L. (2010). Authoritative knowledge, the technological imperative and women's responses to prenatal diagnostic technologies. Culture, Medicine, and Psychiatry, 34(4), 590-614.

Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf

Mills, C. B., & Erzikova, E. (2012). Prenatal Testing, Disability, and Termination: An Examination of Newspaper Framing. Disability Studies Quarterly, 32(3).

Moynihan, R., Bero, L., Ross-Degnan, D., Henry, D., Lee, K., Watkins, J., . . . Soumerai, S. B. (2000). Coverage by the news media of the benefits and risks of medications. New England Journal of Medicine, 342(22), 1645-1650.

Moynihan, R., Heath, I., & Henry, D. (2002). Selling sickness: the pharmaceutical industry and disease mongeringCommentary: Medicalisation of risk factors. Bmj, 324(7342), 886-891.

Natoli, J. L., Ackerman, D. L., McDermott, S., & Edwards, J. G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). Prenatal diagnosis, 32(2), 142-153.

Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genetics in Medicine, 13(11), 913-920.

Pew Research Center. (2010a). Americans Spending More Time Following the News. Retrieved 08-26, 2014, from http://www.people-press.org/files/legacy-pdf/652.pdf

Pew Research Center. (2010c). How News Happens. Retrieved 08-26, 2014, from http://www.journalism.org/2010/01/11/how-news-happens/

Pew Research Center. (2012). Where Americans Get News. Retrieved 08-26, 2014, from http://www.journalism.org/media-indicators/where-americans-get-news/

Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Boland, I. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. Health Expectations, 11(4), 355-365.

President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. (1983). Screening and Counseling for Genetic Conditions: A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs. Washington, DC: U. S. Government Printing Office Retrieved from http://bioethics.georgetown.edu/pcbe/reports/past_commissions/geneticscreening.pdf.

Press, N. A. (2008). Genetic Testing and Screening. From Birth to Death and Bench to Clinic: The Hastings Center Bioethics Briefing Book for Journalists, Policymakers, and Campaigns, 73-78.

Press, N. A., & Browner, C. H. (1994). Collective silences, collective fictions. Women and Prenatal Testing: Facing the Challenges of Genetic Technology, 209-210.

Press, N. A., & Browner, C. H. (1997a). Why Women Say Yes to Prenatal Diagnosis. Social Science and Medicine, 45(7), 979-989.

Press, N. A., & Browner, C. H. (1997c). Why women say yes to prenatal diagnosis. Social Science & Medicine, 45(7), 979-989.

Rapp, R. (1999). Testing women, testing the fetus: The social impact of amniocentesis in America.
Riley, C. A. (2005). Disability and the media: prescriptions for change. Hanover, NH: University Press of New England.
Saxton, M. (2010). Disability rights and selective abortion. In L. J. Davis (Ed.), The Disability Studies Reader (3 ed., pp. 120-132). New York, NY: Routledge.
Schwitzer, G. (2008). How do US journalists cover treatments, tests, products, and procedures? An evaluation of 500 stories. PLoS medicine, 5(5), e95.
Scully, J. L. (2008). Disability and genetics in the era of genomic medicine. Nature Reviews Genetics, 9(10), 797-802.
Seavilleklein, V. (2009). Challenging the rhetoric of choice in prenatal screening. Bioethics, 23(1), 68-77.
Shapiro, J. P. (1993). No pity : people with disabilities forging a new civil rights movement. New York: Times Books.
Shuchman, M., & Wilkes, M. S. (1997). Medical scientists and health news reporting: a case of miscommunication. Ann Intern Med, 126(12), 976-982.
St-Jacques, S., Grenier, S., Charland, M., Forest, J. C., Rousseau, F., & Légaré, F. (2008). Decisional needs assessment regarding Down syndrome prenatal testing: a systematic review of the perceptions of women, their partners and health professionals. Prenatal diagnosis, 28(13), 1183-1203.
Taylor, A. (2013). " Lives Worth Living:" Theorizing Moral Status and Expressions of Human Life. Disability Studies Quarterly, 33(4).
The Noninvasive Prenatal Screening Work Group. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. Genetics in Medicine, 15(5), 395-398.
Tremain, S. (2006). Reproductive Freedom, Self-Regulation, and the Government of Impairment in Utero. Hypatia, 21(1), 35-53. doi: 10.2307/3811076
van den Berg, M., Timmermans, D. R., Ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2005). Are pregnant women making informed choices about prenatal screening? Genet Med, 7(5), 332-338.
van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. Patient Educ Couns, 63(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007
van den Heuvel, A., Chitty, L., Dormandy, E., Newson, A., Deans, Z., Attwood, S., . . . Marteau, T. M. (2010). Will the introduction of non-invasive prenatal diagnostic testing erode informed choices? An experimental study of health care professionals. Patient Educ Couns, 78(1), 24-28. doi: 10.1016/j.pec.2009.05.014
Wilson, A., Bonevski, B., Jones, A., & Henry, D. (2009). Media reporting of health interventions: signs of improvement, but major problems persist. PLoS One, 4(3), e4831. e4831.

Chapter 5

ACOG Office of Communications. (2007a). ACOG's Screening Guidelines on Chromosomal Abnormalities What They Mean to Patients and Physicians. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr05-07-07-1.cfm

ACOG Office of Communications. (2007c). New Recommendations for Down Syndrome: Screening Should Be Offered to All Pregnant Women. ACOG News Release. Retrieved 23 January, 2011, from acog.org/…/nr01-02-07-1.cfm

Alderson, P. (2001). Down's syndrome: cost, quality and value of life. Social Science & Medicine (1982), 53(5), 627-638.

American College of Obstetricians and Gynecologists. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. Obstetrics and gynecology, 110(6), 1459.

American College of Obstetricians and Gynecologists. (2012). Noninvasive prenatal testing for fetal aneuploidy. Obstet Gynecol, 120(545), 1532–1534.

Americans with Disabilities Act, Pub. L. No. 110-325 (1990).

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. American Journal of Public Health, 89(11), 1649-1657.

Asch, A. (2000). Why I haven't changed my mind about prenatal diagnosis: Reflections and refinements. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.

Asch, A. (2003). Disability equality and prenatal testing: contradictory or compatible? Florida State University Law Review. Florida State University. College Of Law, 30(2), 315-342.

Baily, M. A., & Murray, T. H. (2008). Ethics, evidence, and cost in newborn screening. Hastings Center Report, 38(3), 23-31.

Bakke, A. (2013). Disabling the Rhetoric of Prenatal Genetic Testing: A Critical Analysis of Online Self-Help Literature. Critical Disability Discourse, 5, 11-38.

Ballantyne, A., Newson, A., Luna, F., & Ashcroft, R. (2009). Prenatal diagnosis and abortion for congenital abnormalities: is it ethical to provide one without the other? The American Journal of Bioethics, 9(8), 48-56.

Biesecker, B. B., & Hamby, L. (2000). What difference the disability community arguments should make for the delivery of prenatal genetic information. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.

Bindra, R., Heath, V., Liao, A., Spencer, K., & Nicolaides, K. H. (2002). One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. Ultrasound in Obstetrics and Gynecology, 20(3), 219-225. doi: 10.1046/j.1469-0705.2002.00808.x

Botkin, J. R., Clayton, E. W., Fost, N. C., Burke, W., Murray, T. H., Baily, M. A., . . . Ross, L. F. (2006). Newborn Screening Technology: Proceed With Caution. Pediatrics, 117(5), 1793-1799. doi: 10.1542/peds.2005-2547

Browner, C. H. (2000). Situating Women's Reproductive Activities. American Anthropologist, 102(4), 773-788.

Browner, C. H., & Press, N. (1996). The production of authoritative knowledge in American prenatal care. Medical Anthropology Quarterly, 10(2), 141-156.

Browner, C. H., & Press, N. A. (1995). The normalization of prenatal diagnostic screening. Conceiving the new world order: The global politics of reproduction, 307-322.

Buchanan, A., Brock, D. W., Daniels, N., & Wikler, D. (2000). From Chance to Choice: Genetics and Justice. Cambridge: Cambridge Univ Pr.

Burke, W., Atkins, D., Gwinn, M., Guttmacher, A., Haddow, J., Lau, J., . . . Wiesner, G. L. (2002). Genetic Test Evaluation: Information Needs of Clinicians, Policy Makers, and the Public. American Journal of Epidemiology, 156(4), 311-318. doi: 10.1093/aje/kwf055

Chao, S., Roberts, J. S., Marteau, T. M., Silliman, R., Cupples, L. A., & Green, R. C. (2008). Health behavior changes after genetic risk assessment for Alzheimer disease: The REVEAL Study. Alzheimer disease and associated disorders, 22(1).

Childress, J. F., Faden, R. R., Gaare, R. D., Gostin, L. O., Kahn, J., Bonnie, R. J., . . . Nieburg, P. (2002). Public health ethics: mapping the terrain. The Journal of Law, Medicine & Ethics, 30(2), 170-178.

Chou, W.-Y. S., Hunt, Y., Folkers, A., & Augustson, E. (2011). Cancer Survivorship in the Age of YouTube and Social Media: A Narrative Analysis. J Med Internet Res, 13(1), e7. doi: 10.2196/jmir.1569

Clerici, C. A., Veneroni, L., Bisogno, G., Trapuzzano, A., & Ferrari, A. (2012). Videos on rhabdomyosarcoma on YouTube: an example of the availability of information on pediatric tumors on the web. Journal of pediatric hematology oncology, 34(8), 329-331.

Constantin, C. M., Faucett, A., & Lubin, I. M. (2005). A primer on genetic testing. Journal of Midwifery & Women's Health, 50(3), 197-204.

Cushing, P., & Smith, T. (2009). Multinational Review of English Language Disability Studies Degrees and Courses. Disability Studies Quarterly, 29(3).

Davis, D. S. (2010). Genetic dilemmas : reproductive technology, parental choices, and children's futures. Oxford; New York: Oxford University Press.

Eddens, K. S., Kreuter, M. W., Morgan, J. C., Beatty, K. E., Jasim, S. A., Garibay, L., . . . Jupka, K. A. (2009). Disparities by race and ethnicity in cancer survivor stories available on the web. J Med Internet Res, 11(4), e50. doi: 10.2196/jmir.1163

Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. American Journal of Obstetrics and Gynecology, 191(3), 1044-1048.

Ellis, K. (2015). Disability and Spreadable Media: Access, Representation and Inspiration PornDisability in Popular Culture: Focusing Passion, Creating Community and Expressing Defiance (pp. 139-158): Ashgate Publishing Limited.

Farrell, R., Hawkins, A., Barragan, D., Hudgins, L., & Taylor, J. (2015). Knowledge, understanding and uptake of non-invasive prenatal testing among Latina women. Prenatal diagnosis, n/a-n/a. doi: 10.1002/pd.4599

Fox, S. (2012). Pew Internet: Health. Retrieved 24 January, 2013, from http://www.pewinternet.org/Commentary/2011/November/Pew-Internet-Health.aspx

Fox, S. (2013). Social media in medicine. The Who, What, Where, When & Why of Health Care social media. https://youtu.be/QbP9DtDjBRk: Albert Einstein College of Medicine.

Fox, S., & Duggan, M. (2013). Health online. Pew Research Center's Internet and American life Project.

Gabarron, E., Fernandez-Luque, L., Armayones, M., & Lau, A. Y. S. (2013). Identifying Measures Used for Assessing Quality of YouTube Videos with Patient Health Information: A Review of Current Literature. Interactive Journal of Medical Research, 2(1), e6. doi: 10.2196/ijmr.2465

Gee, J. P. (2010). An introduction to discourse analysis : theory and method. New York [etc.]: Routledge.

Ginsburg, F., & Rapp, R. (1999). Fetal reflections: Confessions of two feminist anthropologists as mutual informats. In L. M. Morgan & M. W. Michaels (Eds.), Fetal subjects, feminist positions. Philadelphia: University of Pennsylvania Press.

Green, R. C., Roberts, J. S., Cupples, L. A., Relkin, N. R., Whitehouse, P. J., Brown, T., . . . Group, R. S. (2009). Disclosure of APOE genotype for risk of Alzheimer's disease. The New England journal of medicine, 361(3), 245-254.
Groce, N. E. (1985). Everyone here spoke sign language : hereditary deafness on Martha's Vineyard. Cambridge, Mass.: Harvard University Press.
Haller, B. A. (2010). Representing disability in an ableist world : essays on mass media. Louisville, KY: The Advocado Press.
Harris, A., Kelly, S. E., & Wyatt, S. (2014). Autobiologies on YouTube: Narratives of Direct-to-Consumer Genetic Testing. New Genet Soc, 33(1), 60-78. doi: 10.1080/14636778.2014.884456
Hiatt, H. H. (1975). Protecting the medical commons: who is responsible? The New England journal of medicine, 293(5), 235-241. doi: 10.1056/nejm197507312930506
Hsieh, H., & Shannon, S. (2005). Three approaches to qualitative content analysis. Qual Health Res, 15(9), 1277-1288. doi: 10.1177/1049732305276687
Hughes, B., Joshi, I., & Wareham, J. (2008). Health 2.0 and Medicine 2.0: Tensions and Controversies in the Field. J Med Internet Res, 10(3), e23. doi: 10.2196/jmir.1056
Ivry, T. (2008). At the Back Stage of Prenatal Care: Japanese Ob-Gyns Negotiating Prenatal Diagnosis. Medical Anthropology Quarterly, 20(4), 441-468.
Jaarsma, P., & Welin, S. (2012). Autism as a natural human variation: Reflections on the claims of the neurodiversity movement. Health Care Analysis, 20(1), 20-30.
Kalfoglou, A., Suthers, K., Scott, J., & Hudson, K. (2004). Reproductive genetic testing: What America thinks. Washington, DC: Genetics and Public Policy Center.
Kellogg, G., Slattery, L., Hudgins, L., & Ormond, K. (2014). Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing. Journal of genetic counseling, 23(5), 805-813. doi: 10.1007/s10897-014-9694-7
Kelly, S., & Farrimond, H. (2012). Non-invasive prenatal genetic testing: a study of public attitudes. Public Health Genomics, 15(2), 73-81.
Khoury, M., McBride, C., Schully, S., Ioannidis, J. P., Feero, W. G., Janssens, A. C. J., . . . Cargill, M. (2009). The scientific foundation for personal genomics: recommendations from a National Institutes of Health–Centers for Disease Control and Prevention multidisciplinary workshop. Genetics in Medicine, 11(8), 559.
Khoury, M. J., Jones, K., & Grosse, S. D. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. Genetics in Medicine, 8(3), 191.
Kittay, E. F., & Kittay, L. (2000). On the expressivity and ethics of selective abortion for disability: conversations with my son. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.
Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Murray, J. C. (2012). Noninvasive whole-genome sequencing of a human fetus. Science translational medicine, 4(137), 137ra176-137ra176.
Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Shendure, J. (2012). Noninvasive whole-genome sequencing of a human fetus. Sci Transl Med, 4(137), 137ra176. doi: 10.1126/scitranslmed.3004323
Klein, D. A. (2011). Medical disparagement of the disability experience: Empirical evidence for the "expressivist objection". AJOB Primary Research, 2(2), 8-20.
Kuppermann, M., Pena, S., Bishop, J. T., Nakagawa, S., Gregorich, S. E., Sit, A., . . . Norton, M. E. (2015). Effect of Enhanced Information, Values Clarification, and Removal of Financial

Barriers on Use of Prenatal Genetic Testing. Obstetrical & Gynecological Survey Obstetrical & Gynecological Survey, 70(1), 7-9.
LaRusse, S., Roberts, J. S., Marteau, T. M., Katzen, H., Linnenbringer, E. L., Barber, M., . . . Relkin, N. R. (2005). Genetic susceptibility testing versus family history-based risk assessment: Impact on perceived risk of Alzheimer disease. Genetics in Medicine, 7(1), 48-53.
Lawson, K. L., Carlson, K., & Shynkaruk, J. M. (2012). The Portrayal of Down Syndrome in Prenatal Screening Information Pamphlets. J Obstet Gynaecol Can, 34(8), 760-768.
Learman, L. A., Kuppermann, M., Gates, E., Nease, R. F., Gildengorin, V., & Washington, A. E. (2003). Social and familial context of prenatal genetic testing decisions: Are there racial/ethnic differences? American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 119C(1), 19-26. doi: 10.1002/ajmg.c.10004
LeGreco, M., & Tracy, S. J. (2009). Discourse tracing as qualitative practice. Qualitative Inquiry, 15(9), 1516-1543.
Leung, T. Y., Chan, L. W., Law, L. W., Sahota, D. S., Fung, T. Y., Leung, T. N., & Lau, T. K. (2009). First trimester combined screening for Trisomy 21 in Hong Kong: outcome of the first 10,000 cases. The Journal of Maternal-Fetal & Neonatal Medicine, 22(4), 300-304. doi: 10.1080/14767050802430842
Lewis, C., Silcock, C., & Chitty, L. S. (2013). Non-invasive prenatal testing for Down's syndrome: pregnant women's views and likely uptake. Public Health Genomics, 16(5), 223-232. doi: 10.1159/000353523
Lippman, A. (1999). Embodied knowledge and making sense of prenatal diagnosis. Journal of genetic counseling, 8(5), 255-274.
Lippman, A., & Wilfond, B. S. (1992). Twice-told tales: stories about genetic disorders. American Journal of Human Genetics, 51(4), 936.
Lo, Y., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W., & Wainscoat, J. S. (1997). Presence of fetal DNA in maternal plasma and serum. the lancet, 350(9076), 485-487.
Lo, Y., Tein, M., Lau, T., Haines, C., Leung, T., Poon, P., . . . Hjelm, N. (1998). Quantitative analysis of fetal DNA in maternal plasma and serum: implications for noninvasive prenatal diagnosis. The American Journal of Human Genetics, 62(4), 768-775.
Lupton, D. (2014). Critical Perspectives on Digital Health Technologies. Sociology compass, 8(12), 1344-1359.
Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: a commentary. American journal of medical genetics. Part A, 155A(8), 1777-1785.
Malone, F. D., Canick, J. A., Ball, R. H., Nyberg, D. A., Comstock, C. H., Bukowski, R., . . . D'Alton, M. E. (2005). First-Trimester or Second-Trimester Screening, or Both, for Down's Syndrome. New England Journal of Medicine, 353(19), 2001-2011. doi: doi:10.1056/NEJMoa043693
Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. Prenatal diagnosis, 19(9), 808-812. doi: 10.1002/(sici)1097-0223(199909)19:9<808::aid-pd637>3.0.co;2-b
Markel, H. (1997). Scientific advances and social risks: historical perspectives of genetic screening programs for Sickle Cell Disease, Tay-Sachs Disease, neural tube defects and

Down Syndrome, 1970-1997. Medical and Public Health Law. http://biotech.law.lsu.edu/research/fed/tfgt/appendix6.htm
Markens, S., Browner, C. H., & Preloran, M. H. (2010). Interrogating the dynamics between power, knowledge and pregnant bodies in amniocentesis decision making. Sociology of health & illness, 32(1), 37-56.
Markens, S., Browner, C. H., & Press, N. A. (1999). Because of the risks': how US pregnant women account for refusing prenatal screening. Social Science & Medicine, 49(3), 359-369.
Marteau, T., Drake, H., & Bobrow, M. (1994). Counselling following diagnosis of a fetal abnormality: the differing approaches of obstetricians, clinical geneticists, and genetic nurses. Journal of medical genetics, 31(11), 864-867.
Marteau, T., Plenicar, M., & Kidd, J. (1993). Obstetricians presenting amniocentesis to pregnant women: practice observed. Journal of Reproductive and Infant Psychology, 11(1), 3-10.
Mazanderani, F., O'Neill, B., & Powell, J. (2013). "People power" or "pester power"? YouTube as a forum for the generation of evidence and patient advocacy. Patient Educ Couns, 93(3), 420-425. doi: http://dx.doi.org/10.1016/j.pec.2013.06.006
Media Insight Project, Rosenstiel, T., Sonderman, J., Loker, K., Tran, M., Topmpson, T., . . . Junius, D. (2014). The Personal New Cycle. Retrieved 08-25, 2014, from http://www.americanpressinstitute.org/wp-content/uploads/2014/03/The_Media_Insight_Project_The_Personal_News_Cycle_Final.pdf
Miron-Shatz, T., Hanoch, Y., Graef, D., & Sagi, M. (2009). Presentation Format Affects Comprehension and Risk Assessment: The Case of Prenatal Screening. Journal of Health Communication, 14(5), 439-450. doi: 10.1080/10810730903032986
Moorhead, S. A., Hazlett, D. E., Harrison, L., Carroll, J. K., Irwin, A., & Hoving, C. (2013). A New Dimension of Health Care: Systematic Review of the Uses, Benefits, and Limitations of Social Media for Health Communication. Journal of Medical Internet Research, 15(4), e85. doi: 10.2196/jmir.1933
Muller, C., & Cameron, L. D. (2015). It's complicated – Factors predicting decisional conflict in prenatal diagnostic testing. Health Expectations, n/a-n/a. doi: 10.1111/hex.12363
Murkoff, H. E., & Mazel, S. (2008). What to expect when you're expecting (4th ed.). New York: Workman Pub.
Nadler, H. L. (1968). Antenatal detection of hereditary disorders. Pediatrics, 42(6), 912-918.
Naslund, J. A., Grande, S. W., Aschbrenner, K. A., & Elwyn, G. (2014). Naturally occurring peer support through social media: the experiences of individuals with severe mental illness using YouTube. PLoS One, 9(10), e110171. doi: 10.1371/journal.pone.0110171
National Coalition for Health Professional Education in Genetics, & National Society of Genetic Counselors. (2012). Prenatal Screening/Testing Options for Trisomy 21 and Other Fetal Aneuploidies. In N.-I. P. T. N. Factsheet (Ed.).
Nelson, J. L. (1998). The meaning of the act: reflections on the expressive force of reproductive decision making and policies. Kennedy Institute Of Ethics Journal, 8(2), 165-182.
Nicolaides, K. H. (2011). Screening for fetal aneuploidies at 11 to 13 weeks. Prenatal diagnosis, 31(1), 7-15. doi: 10.1002/pd.2637
Nicolaides, K. H., Spencer, K., Avgidou, K., Faiola, S., & Falcon, O. (2005). Multicenter study of first-trimester screening for trisomy 21 in 75 821 pregnancies: results and estimation of

the potential impact of individual risk-orientated two-stage first-trimester screening. Ultrasound in Obstetrics and Gynecology, 25(3), 221-226. doi: 10.1002/uog.1860
Norton, M. E., Brar, H., Weiss, J., Karimi, A., Laurent, L. C., Caughey, A. B., . . . Song, K. (2012). Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18. American Journal of Obstetrics and Gynecology, 207(2), 137.e131-137.e138. doi: http://dx.doi.org/10.1016/j.ajog.2012.05.021
Oliver, M. (1990). The individual and social models of disability. Paper presented at the Joint Workshop of the Living Options Group and the Research Unit of the Royal College of Physicians.
Oliver, M. (1996). Defining Impairment and Disability: Issues at Stake. Paper presented at the Exploring the divide, University of Leeds.
Ormond, K. E., Gill, C. J., Semik, P., & Kirschner, K. L. (2003). Attitudes of health care trainees about genetics and disability: Issues of access, health care communication, and decision making. Journal of Genetic Counseling, 12(4), 333-349.
Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., . . . Canick, J. A. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genetics in Medicine, 13(11), 913-920.
Pilnick, A. (2004). 'It's Just One of the Best Tests that We've Got at the Moment': the Presentation of Nuchal Translucency Screening for Fetal Abnormality in Pregnancy. Discourse & Society, 15(4), 451-465. doi: 10.1177/0957926504043710
Potter, B. K., O'Reilly, N., Etchegary, H., Howley, H., Graham, I. D., Walker, M., . . . Wilson, B. J. (2008). Exploring informed choice in the context of prenatal testing: findings from a qualitative study. Health Expectations, 11(4), 355-365. doi: 10.1111/j.1369-7625.2008.00493.x
Press, N., & Browner, C. (1994). Collective silences, collective fictions. Women and Prenatal Testing: Facing the Challenges of Genetic Technology, 209-210.
Press, N., & Browner, C. H. (1997). Why Women Say Yes to Prenatal Diagnosis. Social Science and Medicine, 45(7), 979-989.
Press, N., Wilfond, B., Murray, M., & Burke, W. (2011). The power of knowledge: How carrier and prenatal screening altered the clinical goals of genetic testing. In W. Burke, K. A. Edwards, S. Goering, S. Holland & S. B. Trinidad (Eds.), Achieving Justice in Genomic Translation: Re-Thinking the Pathway to Benefit: Re-Thinking the Pathway to Benefit: Oxford University Press, USA.
Rapp, R. (1993). Amniocentesis in sociocultural perspective. Journal of genetic counseling, 2(3), 183-196.
Rapp, R. (1996). The Power of "Positive" Diagnosis: Medical and Maternal Discourses on Amniocentesis. In D. Bassin, M. Honey & M. M. Kaplan (Eds.), Representations of motherhood: Yale University Press.
Rapp, R. (1998). Refusing prenatal diagnosis: The meanings of bioscience in a multicultural world. Science, technology & human values, 23(1), 45-70.
Rapp, R. (1999). Testing women, testing the fetus: The social impact of amniocentesis in America.

Reed, K. (2009). 'It's them faulty genes again': women, men and the gendered nature of genetic responsibility in prenatal blood screening. Sociology of health & illness, 31(3), 343-359. doi: 10.1111/j.1467-9566.2008.01134.x
Riley, C. A. (2005). Disability and the media: prescriptions for change. Hanover, NH: University Press of New England.
Roberts, J. S., Barber, M., Brown, T. M., Cupples, L. A., Farrer, L. A., LaRusse, S. A., . . . Green, R. C. (2004). Who seeks genetic susceptibility testing for Alzheimer's disease? Findings from a multisite, randomized clinical trial. Genetics in Medicine, 6(4).
Rothman, B. K. (1994). The Tentative Pregnancy: Then and Now. In K. H. Rothenberg & E. J. Thomson (Eds.), Women and prenatal testing : facing the challenges of genetic technology. Columbus: Ohio State University Press.
Saxton, M. (2006). Disability Rights and Selective Abortion. In L. J. Davis (Ed.), The disability studies reader (2nd ed., pp. xviii, 451 p.). New York: Routledge.
Secretary's Advisory Committee on Genetic Testing. (2000). Enhancing the oversight of genetic tests: recommendations of the SACGT. from http://oba.od.nih.gov/oba/sacgt/highlights/sacgt012700summ.pdf
Shapiro, J. P. (1993). No pity : people with disabilities forging a new civil rights movement. New York: Times Books.
Skotko, B. G. (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? Archives of disease in childhood, 94(11), 823-826.
Snyder, M. W., Simmons, L. E., Kitzman, J. O., Coe, B. P., Henson, J. M., Daza, R. M., . . . Gammill, H. S. Copy-number variation and false positive prenatal aneuploidy screening results. New England Journal of Medicine.
Spencer, K., Souter, V., Tul, N., Snijders, R., & Nicolaides, K. H. (1999). A screening program for trisomy 21 at 10–14 weeks using fetal nuchal translucency, maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A. Ultrasound in Obstetrics and Gynecology, 13(4), 231-237. doi: 10.1046/j.1469-0705.1999.13040231.x
Stapleton, H., Kirkham, M., & Thomas, G. (2002). Qualitative study of evidence based leaflets in maternity care. BMJ : British Medical Journal, 324(7338), 639-639.
Steinbock, B. (2000). Disability, Prenatal Testing, and Selective Abortion. In E. Parens & A. Asch (Eds.), Prenatal testing and disability rights: Georgetown Univ Press.
Stern, A. M. (2005). Sterilized in the name of public health: race, immigration, and reproductive control in modern California. American Journal of Public Health, 95(7), 1128.
Syed-Abdul, S., Fernandez-Luque, L., Jian, W.-S., Li, Y.-C., Crain, S., Hsu, M.-H., . . . Liou, D.-M. (2013). Misleading Health-Related Information Promoted Through Video-Based Social Media: Anorexia on YouTube. J Med Internet Res, 15(2), e30. doi: 10.2196/jmir.2237
Task Force on Genetic Testing, Holtzman, N. A., & Watson, M. S. (1998). Promoting safe and effective genetic testing in the United States : final report of the Task Force on Genetic Testing. Baltimore: Johns Hopkins University Press.
Taylor, J. S. (2000). Of sonograms and baby prams: Prenatal diagnosis, pregnancy, and consumption. Feminist Studies, 26(2), 391-418.
Ten Great Public Health Achievements--United States, 1900-1999. (1999). JAMA: The Journal of the American Medical Association, 281(16), 1481-1481. doi: 10.1001/jama.281.16.1481
Thomas, K., Bourke, J., Girdler, S., Bebbington, A., Jacoby, P., & Leonard, H. (2011). Variation Over Time in Medical Conditions and Health Service Utilization of Children with Down

Syndrome. The Journal of Pediatrics, 158(2), 194-200.e191. doi: http://dx.doi.org/10.1016/j.jpeds.2010.08.045
US Department of Energy Genome Programs. (2011). Human Genome Project Information from http://web.ornl.gov/sci/techresources/Human_Genome/index.shtml
van den Berg, M., Timmermans, D. R., ten Kate, L. P., van Vugt, J. M., & van der Wal, G. (2006). Informed decision making in the context of prenatal screening. Patient Educ Couns, 63(1-2), 110-117. doi: 10.1016/j.pec.2005.09.007
Vanzwieten, M., Willems, D., Knegt, L., & Leschot, N. (2006). Communication with patients during the prenatal testing procedure: An explorative qualitative study. Patient Education and Counseling, 63(1-2), 161-168. doi: 10.1016/j.pec.2005.09.013
Wald, N. J., Huttly, W. J., & Hackshaw, A. K. (2003). Antenatal screening for Down's syndrome with the quadruple test. The Lancet, 361(9360), 835-836. doi: http://dx.doi.org/10.1016/S0140-6736(03)12680-3
Wald, N. J., Huttly, W. J., Murphy, K. W., K, A., Bestwick, J. P., & Rodeck, C. H. (2009). Antenatal screening for Down's syndrome using the Integrated test at two London hospitals. Journal of Medical Screening, 16(1), 7-10. doi: 10.1258/jms.2009.008094
Yang, M., Seo, J., Patel, A., & Sansgiry, S. S. (2012). Content Analysis of the Videos Featuring Prescription Drug Advertisements in Social Media: YouTube. Drug Information Journal, 46(6), 715-722. doi: 10.1177/0092861512462020
Zick, C. D., Mathews, C. J., Roberts, J. S., Cook-Deegan, R., Pokorski, R. J., & Green, R. C. (2005). Genetic Testing For Alzheimer's Disease And Its Impact On Insurance Purchasing Behavior. Health Affairs, 24(2), 483-490. doi: 10.1377/hlthaff.24.2.483
Zimmern, R. L., & Kroese, M. (2007). The evaluation of genetic tests. Journal of Public Health, 29(3), 246-250. doi: 10.1093/pubmed/fdm028

Chapter 6

American College of Obstetricians and Gynecologists. (2007). Screening for Fetal Chromosomal Abnormalities. Obstetrics & Gynecology, 109(1).
American College of Obstetricians Gynecologists. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. Obstetrics and gynecology, 110(6), 1459.
Benn, P. A., Egan, J. F., Fang, M., & Smith-Bindman, R. (2004). Changes in the utilization of prenatal diagnosis. Obstet Gynecol, 103(6), 1255-1260. doi: 10.1097/01.aog.0000127008.14792.14
Benn, P. A., Horne, D., Briganti, S., & Greenstein, R. M. (1995). Prenatal diagnosis of diverse chromosome abnormalities in a population of patients identified by triple-marker testing as screen positive for Down syndrome. Am J Obstet Gynecol, 173(2), 496-501.
Besser, L. M., Shin, M., Kucik, J. E., & Correa, A. (2007). Prevalence of down syndrome among children and adolescents in metropolitan Atlanta. Birth Defects Res A Clin Mol Teratol, 79(11), 765-774. doi: 10.1002/bdra.20404
Bishop, J., Huether, C. A., Torfs, C., Lorey, F., & Deddens, J. (1997). Epidemiologic study of Down syndrome in a racially diverse California population, 1989-1991. Am J Epidemiol, 145(2), 134-147.
Bornstein, E., Lenchner, E., Donnenfeld, A., Barnhard, Y., Seubert, D., & Divon, M. Y. (2009). Advanced maternal age as a sole indication for genetic amniocentesis; risk-benefit analysis

based on a large database reflecting the current common practice. J Perinat Med, 37(2), 99-102. doi: 10.1515/jpm.2009.032

Caruso, T. M., Westgate, M. N., & Holmes, L. B. (1998). Impact of prenatal screening on the birth status of fetuses with Down syndrome at an urban hospital, 1972-1994. Genet Med, 1(1), 22-28. doi: 10.1097/00125817-199811000-00006

Centers for Disease Control and Prevention. (1994). Down syndrome prevalence at birth--United States, 1983-1990. MMWR Morb Mortal Wkly Rep, 43(33), 617-622.

Cocchi, G., Gualdi, S., Bower, C., Halliday, J., Jonsson, B., Myrelid, A., . . . Anneren, G. (2010). International trends of Down syndrome 1993-2004: Births in relation to maternal age and terminations of pregnancies. Birth Defects Res A Clin Mol Teratol, 88(6), 474-479. doi: 10.1002/bdra.20666

Collins, J. S., Olson, R. L., DuPont, B. R., Wolff, D. J., Best, R. G., & Stevenson, R. E. (2002). Prevalence of aneuploidies in South Carolina in the 1990s. Genet Med, 4(3), 131-135. doi: 10.109700125817-200205000-00006

Correa-Villaseñor, A., Cragan, J., Kucik, J., O'Leary, L., Siffel, C., & Williams, L. (2003). The metropolitan Atlanta congenital defects program: 35 years of birth defects surveillance at the Centers for Disease Control and Prevention. Birth Defects Research Part A: Clinical and Molecular Teratology, 67(9), 617-624.

Dzurova, D., & Pikhart, H. (2005). Down syndrome, paternal age and education: comparison of California and the Czech Republic. BMC Public Health, 5, 69. doi: 10.1186/1471-2458-5-69

Egan, J. F., Benn, P. A., Zelop, C. M., Bolnick, A., Gianferrari, E., & Borgida, A. F. (2004). Down syndrome births in the United States from 1989 to 2001. Am J Obstet Gynecol, 191(3), 1044-1048. doi: 10.1016/j.ajog.2004.06.050

Egan, J. F., Smith, K., Timms, D., Bolnick, J. M., Campbell, W. A., & Benn, P. A. (2011). Demographic differences in Down syndrome livebirths in the US from 1989 to 2006. Prenat Diagn, 31(4), 389-394. doi: 10.1002/pd.2702

Forrester, M. B., & Merz, R. D. (1999). Prenatal diagnosis and elective termination of Down syndrome in a racially mixed population in Hawaii, 1987-1996. Prenat Diagn, 19(2), 136-141.

Forrester, M. B., & Merz, R. D. (2002). Epidemiology of Down syndrome (Trisomy 21), Hawaii, 1986-97. Teratology, 65(5), 207-212. doi: 10.1002/tera.10042

Forrester, M. B., & Merz, R. D. (2003). Maternal age-specific Down syndrome rates by maternal race/ethnicity, Hawaii, 1986-2000. Birth Defects Res A Clin Mol Teratol, 67(9), 625-629. doi: 10.1002/bdra.10112

Gordis, L. (2009). Epidemiology. Philadelphia: Elsevier/Saunders.

Hahn, J. A., & Shaw, G. M. (1993). Trends in Down's syndrome prevalence in California, 1983-1988. Paediatr Perinat Epidemiol, 7(4), 450-460.

Henry, G. P., Britt, D. W., & Evans, M. I. (2008). Screening advances and diagnostic choice: the problem of residual risk. Fetal Diagn Ther, 23(4), 308-315. doi: 10.1159/000123619

Huether, C. A., Haroldson, K., Ellis, P. M., & Ramsay, C. N. (1996). Impact of prenatal diagnosis on revised livebirth prevalence estimates of Down syndrome in the Lothian region of Scotland, 1978-1992. Genet Epidemiol, 13(4), 367-375. doi: 10.1002/(SICI)1098-2272(1996)13:4<367::AID-GEPI5>3.0.CO;2-1

James, L. M. (1993). Maps of birth defects occurrence in the U.S., Birth Defects Monitoring Program (BDMP)/CPHA, 1970-1987. Teratology, 48(6), 551-646. doi: 10.1002/tera.1420480605

Krivchenia, E., Huether, C. A., Edmonds, L. D., May, D. S., & Guckenberger, S. (1993). Comparative epidemiology of Down syndrome in two United States population, 1970-1989. Am J Epidemiol, 137(8), 815-828.

Mansfield, C., Hopfer, S., & Marteau, T. M. (1999). Termination rates after prenatal diagnosis of Down syndrome, spina bifida, anencephaly, and Turner and Klinefelter syndromes: a systematic literature review. Prenatal diagnosis, 19(9), 808-812.

Mason, C. A., Kirby, R. S., Sever, L. E., & Langlois, P. H. (2005). Prevalence is the preferred measure of frequency of birth defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 73(10), 690-692.

McDermott, H., & Johnson, J. L. (2011). Is the prevalence of Down syndrome births in Hawai'i increasing? Hawaii Med J, 70(4), 72-76.

Nadler, H. L. (1968). Antenatal detection of hereditary disorders. Pediatrics, 42(6), 912-918.

National Birth Defects Prevention Network. (2004). Guidelines for Conducting Birth Defects Surveillance L. E. Sever (Ed.) Retrieved from http://www.nbdpn.org/docs/NBDPN_Guidelines2008.pdf

National Center for Health Statistics. (1995-2002). Natality. Retrieved from: http://wonder.cdc.gov/natality.html

National Center for Health Statistics. (2003-2006). Natality. Retrieved from: http://wonder.cdc.gov/natality.html

National Center for Health Statistics. (2007-2012). Natality. Retrieved from: http://wonder.cdc.gov/natality.html

Natoli, J. L., Ackerman, D. L., McDermott, S., & Edwards, J. G. (2012). Prenatal diagnosis of Down syndrome: a systematic review of termination rates (1995–2011). Prenatal diagnosis, 32(2), 142-153.

Olsen, C. L., Cross, P. K., & Gensburg, L. J. (2003). Down syndrome: interaction between culture, demography, and biology in determining the prevalence of a genetic trait. Hum Biol, 75(4), 503-520.

Olsen, C. L., Cross, P. K., Gensburg, L. J., & Hughes, J. P. (1996). The effects of prenatal diagnosis, population ageing, and changing fertility rates on the live birth prevalence of Down syndrome in New York State, 1983-1992. Prenat Diagn, 16(11), 991-1002. doi: 10.1002/(sici)1097-0223(199611)16:11<991::aid-pd977>3.0.co;2-5

Orton, H., Rickard, R., & Miller, L. (2001). Using active medical record review and capture-recapture methods to investigate the prevalence of Down Syndrome among live-born infants in Colorado. Teratology, 64 Suppl 1, S14-19. doi: 10.1002/tera.1079

Saha, S., Chant, D., & McGrath, J. (2008). Meta-analyses of the incidence and prevalence of schizophrenia: conceptual and methodological issues. International Journal of Methods in Psychiatric Research, 17(1), 55-61. doi: 10.1002/mpr.240

Shin, M., Besser, L. M., Kucik, J. E., Lu, C., Siffel, C., & Correa, A. (2009). Prevalence of Down syndrome among children and adolescents in 10 regions of the United States. Pediatrics, 124(6), 1565-1571. doi: 10.1542/peds.2009-0745

Siffel, C., Correa, A., Cragan, J., & Alverson, C. J. (2004). Prenatal diagnosis, pregnancy terminations and prevalence of Down syndrome in Atlanta. Birth Defects Res A Clin Mol Teratol, 70(9), 565-571. doi: 10.1002/bdra.20064

Vendola, C., Canfield, M., Daiger, S. P., Gambello, M., Hashmi, S. S., King, T., . . . Hecht, J. T. (2010). Survival of Texas infants born with trisomies 21, 18, and 13. Am J Med Genet A, 152A(2), 360-366. doi: 10.1002/ajmg.a.33156

Wilson, M. G., Chan, L. S., & Herbert, W. S. (1992). Birth prevalence of Down syndrome in a predominantly Latino population: a 15-year study. Teratology, 45(3), 285-292. doi: 10.1002/tera.1420450307

Chapter 7

Asch, A. (1999). Prenatal diagnosis and selective abortion: a challenge to practice and policy. American Journal of Public Health, 89(11), 1649-1657.

Hiatt, H. H. (1975). Protecting the medical commons: who is responsible? The New England journal of medicine, 293(5), 235-241. doi: 10.1056/nejm197507312930506

Kitzman, J. O., Snyder, M. W., Ventura, M., Lewis, A. P., Qiu, R., Simmons, L. E., . . . Shendure, J. (2012). Noninvasive whole-genome sequencing of a human fetus. Sci Transl Med, 4(137), 137ra176. doi: 10.1126/scitranslmed.3004323

Snyder, M. W., Simmons, L. E., Kitzman, J. O., Coe, B. P., Henson, J. M., Daza, R. M., . . . Gammill, H. S. Copy-number variation and false positive prenatal aneuploidy screening results. New England Journal of Medicine.

Thomas, K., Bourke, J., Girdler, S., Bebbington, A., Jacoby, P., & Leonard, H. (2011). Variation Over Time in Medical Conditions and Health Service Utilization of Children with Down Syndrome. The Journal of Pediatrics, 158(2), 194-200.e191. doi: http://dx.doi.org/10.1016/j.jpeds.2010.08.045

VITA

Anjali Truitt earned her Bachelor of Science in Biology and a second major in Individualized Studies in Liberal Arts from Loyola Marymount University in Los Angeles, California. She completed her Master of Public Health in Community Health Sciences at the University of Illinois at Chicago. During this training, she participated in the Illinois Leadership Education in Neurodevelopmental and Related Disabilities (LEND) traineeship, specializing in autism spectrum disorders. In 2015, she finished her Doctor of Philosophy in Public Health Genetics at the University of Washington.