# Preimplantation genetic testing and perceptions of disability: A content analysis

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## Abstract

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Preimplantation genetic testing (PGT) is a technique used in tandem with in vitro fertilization (IVF) to identify embryos free of genetic conditions for later implantation. Hundreds of genetic conditions can be tested for though PGT, such as down syndrome, cystic fibrosis, and Duchenne muscular dystrophy. The overall purpose of this study is to understand how disability arising from genetic conditions is characterized in clinical communications surrounding the use of PGT. The specific research questions are to (1) understand how disability is portrayed in online patient-facing PGT texts from fertility clinics and (2) determine if these texts show bias regarding whether or not a patient should have PGT, or raise a child with a disability. Texts (N = 33) were analyzed via content analysis to fill the knowledge gap surrounding how these texts contribute to disability discourse. Two major themes emerged from the data: the importance of normal bodies and a fear of disability. These themes are supported by previous literature, and illustrate that texts portray people with disabilities inaccurately and show bias in relation to disability. The discussion explores these themes in more depth and considers next steps for research, recommendations for PGT texts, and limitations of the study.

# I. INTRODUCTION

Preimplantation genetic testing (PGT) is a technique used in tandem with in vitro fertilization (IVF) in order to identify embryos free of genetic conditions for later implantation. PGT is typically offered by IVF clinicians to patients who are likely to pass on a genetic condition or have had previous unexplained miscarriages. Before implantation, embryos are tested for certain genetic mutations and only embryos free of those sequences are eligible for transfer to the uterus.[2] This technique has been successfully used in humans since the early 2000's, and the list of traits it can be used for continues to grow in the present day.[10, 30]

Currently, PGT is able to test embryos for single gene conditions such as cystic fibrosis or achondroplasia, sex-linked conditions like Duchenne muscular dystrophy, and conditions related to aneuploidy such as Down syndrome or some intersex conditions.[16, 20] PGT is also used to select embryos for characteristics such as sex or human leukocyte antigen (HLA) type. Many other conditions are available for testing as well, such as forms of deafness, forms of blindness, early onset Alzheimer's disease, and early onset Parkinson's disease.[20] In some cases, people choose to implant embryos that have certain conditions such as deafness.[22] PGT can help people experiencing multiple miscarriages carry a pregnancy to term, or PGT can be used to prevent a pregnancy resulting in a child with a genetic condition, whether or not that condition is acutely fatal. An estimated 4-6% of IVF cycles employ PGT each year.[36]

Legal restrictions on PGT are virtually non-existent in the United States (US).[7] While the US is able to screen for all of the same conditions as countries that regulate PGT more closely, federal regulation lags behind other countries due to the lack of a national health care system in the US, disagreements over which federal agency should be charged with regulation of PGT, and

the proximity of PGT to debates surrounding abortion.[7] This is leaves regulation up to individual states, but no states have laws specifically applying to PGT; rather states tend to regulate the use of IVF and the treatment of unused embryos.[7] Providers of PGT must adhere to these state laws along with other medical laws such as informed consent, however, who can use PGT or ways in which it can be used remains unregulated. Provider discretion determines what conditions are tested for and for what reasons testing would be offered.

With little regulatory oversight for PGT, many ethical and social dilemmas related to PGT's use emerge. One of these issues lies in PGT's relationship with disability communities. This form of genetic testing has been seen by disability activists and ethicists as a tool for modern eugenics, as selecting who should have a chance at life based on their genetic makeup is at the root of PGT, whether that is its intent or its consequence.[1] The expressivist objection, typically used to argue against the use of prenatal testing, can be applied equally well to PGT. It argues that this kind of testing expresses disvalue for people living with the conditions tested for and is therefore harmful to these individuals and their communities.[14, 25, 29] Disability rights activists also understand that the selection against certain disabilities through the use of PGT could lead to differential treatment and erasure of those with disabilities.[4, 9, 26, 33] In addition, PGT reinforces the medical model of disability which is historically paternalistic, coercive, and typically rejected by proponents of disability rights in favor of the social model.[18, 26]

Medicine, disability, and language have been inextricably linked for centuries. Medical institutions have historically used value-laden language with the purpose of controlling and manipulating people with disabilities in order to marginalize them and remove them from society.[6, 24, 26, 31, 35] Words like *feebleminded*, *abnormal*, *defective*, and *unfit* helped to distance the disabled population from the non-disabled population, making the United States' eugenics

movement of the early 1900's and mass sterilizations of people with disabilities throughout the 20th century more palatable to the general public.[6, 8, 11, 19, 24, 32] Many of these words and other ableist terms are still used today in medical texts when describing people with disabilities and their conditions.[27]

With no legal restrictions on the use of PGT in the United States, the rapid expansion of genetic understanding, and the power that medical institutions wield with language, it is more important than ever to understand how fertility clinics are talking about PGT and by extension disability; this could affect perception of disabilities, shape beliefs, and influence parent's decisions regarding testing and implantation. One way to assess how fertility clinics portray disability is through their patient-facing information on PGT. Fertility clinic websites often provide information on PGT, and studies show that patients rely on online medical information when making their own medical decisions and forming opinions on care which could include forming views on disability, genetic testing, and raising children with disabilities.[12, 17, 21] Despite this, no studies have looked at how PGT texts represent disability. As fertility clinics are an authority on reproductive genetics, it is crucial to understand how PGT and disability are portrayed by patient-facing texts from these organizations.

The aims for this study are to (1) understand how disability is portrayed in online patient-facing fertility clinic PGT texts and (2) determine if these texts show bias regarding disability. The paper closes with a discussion of next steps for research in this area and recommendations for inclusive PGT texts, in hopes that people accessing IVF and PGT will be able to make more informed decisions.

# II. METHODS

## Design

This study used cross-sectional qualitative content analysis to answer the research aims outlined above. Filling the knowledge gap surrounding how these texts discuss disability and contribute to disability discourse was the purpose of analyzing these patient-facing clinical texts on PGT from fertility clinics in the north west United States. The analysis process was adapted from LeGreco and Tracy's Discourse Tracing as Qualitative Practice.[23] Content analysis is the most appropriate method to answer the research questions as it allows for analysis of words and language, and takes into account power structures and how they can affect discourse. This is especially relevant when looking at disability from the perspective of the medical institutions, as medical institutions often have good intentions but end up using their authority to perpetuate ableism and dismiss the lived experiences of those living with disabilities.[26, 27]

## My positionality

I am a white, nondisabled, cisgender, hearing female who only speaks and reads English. I am new to the field of disability studies and have a background in psychology and public health genetics. I recognize that I will likely make mistakes as I cannot understand first-hand the varied experiences of people with disabilities. The information and analysis in this paper is based on what I have read, watched, and listened to by disability studies scholars, activists and allies.

## Sampling online texts

A web browser that would not incorporate information from previous searches into current search results was used to identify fertility clinics in the north west United States. Eligibility criteria required clinics to be operating at the time of search, offer PGT, have PGT texts in English, and have an office located in Washington State, Oregon, or Northern California

(conventionally defined as north of San Luis Obispo County). These regions were included as their larger cities (Seattle, Portland, San Jose) where most fertility clinics are located tend to have a similar social-political climate which is fairly progressive. It was thought that progressive cities would have the most progressive texts and therefore would represent the most up-to-date language regarding disability in the context of PGT. Once clinics were identified in these regions, their websites were searched for texts on PGT. Texts were defined as any publicly available information on PGT located on the clinic's website. This could include answers to frequently asked questions, explanations of PGT under clinic services, brochures, or any other kind of educational materials. All texts (N = 33) were archived for consistency between February 8th and February 12th, 2021.

**Analysis**

Data from the fertility clinic texts were analyzed via qualitative coding. A codebook was created *de novo* for the purposes of this study based on a review of varied disability studies literature.[3, 6, 11, 15, 24] All coding and analysis were done in Atlas.ti.[5] Two coders (J.D. and A.S) coded all texts separately. At three points during the coding process, the two coders came together to assure mutual understanding of the codebook and application to the texts. A kappa of 0.73 was calculated, which supports a moderate level of intercoder reliability. After coding was completed, main themes were extracted from the texts via close review of the codes and texts in order to answer the research questions. Main themes were developed by looking at the most frequently used codes and determining what these codes had in common.

# III. RESULTS

**Major themes**

Two major themes emerged from the data: (1) importance of normal bodies and (2) fear of disability. These themes align with previous research in disability discourse and show that patient-facing fertility clinic PGT texts typically portray disability unfavorably and inaccurately, as something that is not within the acceptable norm and something that should be feared and avoided.[28, 29, 34] Each theme is supported and discussed below.

1. Importance of "normal" bodies

This theme emphasizes the idea that there is one "normal" way for a body or mind to be, whether that is on the cellular level or on the visible level, and that way of being is desired. For example, the language that is used to describe genetic conditions or the lack of genetic conditions in these texts is outdated and not inclusive. Examples include *"genetic problem," "abnormal embryos," "genetically perfect," "abnormalities in the baby," "defects," and "difficulties."* Using this kind of value-laden language, instead of opting for more neutral language, misrepresents life with disabilities and encourages the reader to pity and fear those who do not fit the norm.

Other quotes supported the idea that people should only have 46 chromosomes, and other complements were not desired:

> *"[PGT] can help you determine if an embryo has the correct number and combination of chromosomes" (AFA1)*
> *"Every cell in the human body needs to have a total of 46 chromosomes" (CCR1)*
> *"The goal of...testing is to ensure the correct number of chromosomes (46) are present in each embryo" (PNW1)*

Texts often equated genetic conditions with being unhealthy, and being free of genetic conditions with being healthy. Emphasizing this dichotomy ignores the experiences of many people in disability communities with these conditions who are healthy, and presents a misleading and inaccurate image of disability:

> *"[PGT] allows families to make sure that only healthy embryos are transferred back to the intended mother." (SP1)*
> *"[Through our clinic] more than 5,000 healthy babies have been born following the transfer of embryos with the correct number of chromosomes" (CCR1)*
> *"[PGT] is a type of genetic testing that gives you the best chance of having a healthy baby, even if...you carry a genetic disease" (NOVA1)*
> *"If you [and your partner are carriers for a genetic condition], we can use [PGT] to ensure that you do not have a child that is born sick with the disease" (SP1)*

2. Fear of disability

This theme supports the idea that disability should be actively avoided because its occurrence in your family is something to dread. Genetic conditions were often presented as a "risk" and texts implied their occurrence was as similarly disappointing as a miscarriage or inability to conceive.

> *"PGD allows couples to have children without the risk of transmitting the diagnosed disorder to the embryo" (POMA1)*
> *"...any imbalance in the number of chromosomal pairs (aneuploidy) can give rise to a failure of the embryo to implant, cause early pregnancy loss, or other conditions, such as Down's syndrome" (OL2)*
> *"Chromosome count is an important indication of embryo health. Too few or too many chromosomes can lead to a number of issues, such as failure for an embryo to implant, miscarriage, and chromosome abnormalities in a fetus or child, such as Down syndrome, a condition characterized by various significant physical and cognitive difficulties" (PFC1)*

Fear was also communicated through the use of words like “serious” and “severe” to describe genetic conditions or resulting disability, without specifying which conditions were “serious” or life-threatening, leading the reader to believe that most or all conditions tested for are “severe”.

> *“There are diseases that cause death or severe disability in children with two copies of an abnormal gene” (SP1)*
> *“[PGT] offers another choice to carriers of serious genetic disorders and involves making decisions about the future of an affected embryo, rather than an affected pregnancy” (OL2)*
> *“Two parents carrying the same mutation have a 25% chance of having a severely sick baby.” (SP1)*
> *“[PGT gives] us a very good way to identify and prevent some of the most common and terrifying problems…” (NCF1)*

These themes show that online patient-facing PGT texts have negative attitudes regarding disability. These texts tend to portray disability as something unacceptable, outside the norm, unhealthy, unwanted, and something to be fearful of. This portrayal of people with genetic conditions can perpetuate stigma and implicit bias regarding disability, and can prevent patients from making a fully informed decision.

# IV. DISCUSSION

The purpose of this study is to (1) understand how disability is portrayed in online patient-facing fertility clinic PGT texts and (2) determine if these texts show bias regarding disability. Two major themes emerged via the content analysis: the desire to have a “normal” body/mind, and a

fear of disability. The themes indicate that these texts portray disability poorly and are biased against disability as they reinforce the othering of people with genetic conditions. In addition, these results suggest that PGT texts are often written from the perspective of the medical model of disability as opposed to the social model, which tends to be the model preferred by disability activists and scholars, as the texts rely on the medical system to prevent conditions rather than promoting better social and physical environments for people with disabilities.[27] While virtually no research has been done on PGT and disability discourse, previous research on prenatal screening supports that people with disabilities may feel discriminated against by the use of genetic screening technologies, may fear being erased from future generations, and may fear a reduction in research into medical care for their conditions if they become less prevalent.[28, 29, 34] Irresponsible use of PGT could worsen the social climate for those already existing with disabilities, especially if they become a smaller minority as PGT encourages "normality." This could be exacerbated by non-inclusive PGT texts that do not allow readers a chance to gain a more informed view of life with a disability.

The results reveal a few practical changes fertility clinics could make to their PGT texts in order to make them more inclusive and support informed decisions. One of the most impactful changes would be to remove the negative value-laden language. Additionally, including resources from support networks for certain genetic conditions and raising children with disabilities would provide patients with more information on the conditions they are considering testing for. These changes could help remove the implicit bias against those with genetic conditions, decrease the perpetuation of stigma surrounding these conditions, and help patients make a more fully informed decision regarding PGT.

This study was not without its limitations. Fertility clinics in the north west were not as common as originally thought, so the sample size was smaller than anticipated and the regions included had to be expanded. In addition, only texts in English were analyzed, but disability could be portrayed differently in different languages and the difference in portrayal may or may not be similar to English texts. People who read these texts in other languages are still affected by the portrayal of disability and could still have their decision-making impacted. Lastly, texts ranged in relevancy. This made it more difficult to compare texts as some focused on, say, the technical process behind PGT which did not provide very rich data, while other texts focused more on preventing genetic conditions making their data much richer.

Lastly, I wanted to address the language used throughout this paper, as it was chosen thoughtfully. Person-first language was chosen as it is the most inclusive language available, and this form is widely accepted among people with disabilities and major organizations. In addition, person-first language is the University of Washington's official form.[13] However, individual preferences regarding person-first or identity-first language differ, and these individual choices should always be respected. In addition, in an attempt to avoid the use of value-laden language, more neutral words such as "condition" are used instead of words such as "disease" or "disorder" when it comes to genetic conditions. Similarly, I avoided casual use of the word "normal" throughout the paper, as this is a socially defined term that continually changes.[11] Lastly, the word disability is used throughout the paper to cover a multitude of physical and developmental differences between people, and is often used interchangeably with "genetic condition". I recognize that not all disabilities are caused by genetic conditions and not all genetic conditions lead to disabilities, but for the purpose of this paper I am interested in disability-causing genetic conditions.

Further research is needed to understand how prospective patients might perceive disability from these texts, to understand what impact these texts actually have on the decisions of patients, and to highlight additional issues within these texts. People in disability communities who may not be patients should be involved in this research as well in order to discern how language in these texts impacts the populations they describe. The feedback from these participants could further identify issues within texts and generate suggestions for preferred language and resources to include. Finally, expanding this research to other regions in the US, as well as evaluating other languages is an important component for additional research.

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