A Retrospective Qualitative Content Analysis of Parental Perspectives of Child Disability Following Whole Genome Sequencing

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Abstract

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Background: Whole genome sequencing (WGS) is increasingly used to diagnose children with developmental disabilities, but little is known about how it shapes parents' perceptions of disability.
Methods: This qualitative content analysis of 22 parent interviews examined shifts in medical and social model framing across pre-whole genome sequencing, immediately following receipt of results, and at the time of interview.
Results: Prior to WGS, all parents framed their child's disability through concern and uncertainty. Most parents (16/22) remained consistently medical model dominant. A smaller group (6/22) showed perspectives aligned with the social model, either in the immediate post-results period or only at the time of interview.
Discussion: Findings suggest that WGS may reinforce medical model framing for most families, though social model engagement emerges over time for a minority.

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Introduction

I. Developmental and Intellectual Disabilities: Prevalence and Diagnostic Value

Global developmental delay and intellectual disability affect an estimated 1% to 3% of children globally.[1] These children have many different causes for their delays and disabilities, but genetic disorders have been found to be responsible in approximately 50% of cases.[1] Families who receive a diagnosis from genomic sequencing often find it valuable, even if there is no known treatment currently for the disorder.[2] This is because "a genetic diagnosis can lead to positive changes in surveillance, medical management, and access to clinical trials/research,"[2 (p2494)] as well as allowing for potential family planning and access to support groups.

Families with children who have developmental or intellectual disabilities may seek out genetic testing in order to find clear answers to what may be causing their disability. This is despite the potential of receiving a diagnosis that may or may not have known treatment methods. This potential for a diagnosis would hopefully help the families in gaining a better understanding on how to go about medical treatments and support in the future.[3-4] These parents may also seek out genetic testing in order to find the ways they can best support their child at their current stage of development and work to gain access to treatments, whether or not a diagnosis is made.[4]

II. Whole Genome Sequencing (WGS): Varying Findings and Parental Impact

Whole genome sequencing (WGS) findings vary, with some individuals receiving a precise genetic diagnosis, while others do not.[5] The variance of results (explained, partially explained, not explained) may lead to reinforcement of medicalized views, alleviation of guilt, or acceptance and expansion of advocacy for their child.[6] Here, 'medicalized views' means perspectives aligned with the medical model of disability. WGS and shifts in disability perceptions are connected by their causality. By finding out if there is a known genetic cause for their child's disability, it may guide parents toward further medical treatment, non-medical support, or both. Even with a precise genetic diagnosis, treatment is not guaranteed. A diagnosis is often needed for families to access treatments, therapies, and other specialized services, which makes pursuing genetic testing practical for many families. Differing findings from WGS could lead to a shift in a parent's perspective on their child's disability, though in different ways depending on whether the results explained, partially explained, or did not explain the child's disability. Research suggests that parents who receive a genetic diagnosis for their child often focus on treatment and medical management.[6]

An expectation from the author going into this thesis was that if parents find out the cause for their child's disability, whether inherited or not, they may feel relieved or dive into treatment more as an option. However, if their child's disability is not explained in the results, parents may be left with uncertainty or

shifting their focus towards acceptance and advocacy for their child. This is not to suggest that unexplained results directly cause social model thinking, but that uncertainty may open space for perspective shifts.

III. Medical and Social Models of Disability: Frameworks for Understanding Parental Perspectives

These two routes of further pushing for treatment and/or acceptance and advocacy lead directly into two models of disability that directly guided this analysis. The two models that were used as guiding frameworks were the medical and social models of disability. The medical model of disability views disability as an individual issue, where there is a deficit in function that needs to be treated or 'fixed'.[7] The social model, on the other hand, sees disability as a societal issue, one that is imposed by societal barriers such as inaccessible buildings, ableism, and exclusion.[7] This thesis examines whether the framework parents have at the forefront of their minds impacts the ways in which they engage with their child, their treatments, and the disability community as a whole. These differing mindsets may also reinforce harmful thoughts surrounding disability or expand parents' minds about what it means to have a disabled child.

IV. Goals and Objectives

While research in ethical, legal, and social implications (ELSI) of genomic testing, like WGS, in relation to disability are present in the literature,[8-9] as well as studies examining why parents seek out WGS,[3-4,10] no currently existing work has

analyzed parents' shifts in perspective about their child's disability following the receipt of their child's WGS results, while using the medical and social models of disability as a lens. This ultimately leaves a gap in knowledge of how these experiences may change their perspectives over time. This thesis works to address this gap by answering: *How do parents of children with developmental delays retrospectively discuss shifts in perspectives on their child's disability following the receipt of whole genome sequencing results?*

The goal of this thesis is to evaluate the perspectives of parents who have children with developmental delays and have received whole-genome sequencing through the SeqFirst research study. This analysis looked into parental perspectives of their child's disability retrospectively, following the receipt of whole genome sequencing results. This work is important because it helps in gaining a better understanding of how whole genome sequencing may alter parents' views regarding their child's disability regardless of whether or not a diagnosis is made.

Methods

I. Parent Study: SeqFirst DDi

The data for this analysis originated from the Developmental Differences (DDi) arm of the SeqFirst project. In this project, whole-genome sequencing was provided to participating families for free in order to study the impact of providing WGS early in the diagnostic odyssey. The study recruited families who had children under 3 years of age with a moderate to severe developmental delay

or a mild delay plus differences in physical development.[5] They were recruited from Seattle-area early intervention centers or an academic neurodevelopmental clinic from 2021 to 2023 and were randomly assigned to either the sequencing group, who received WGS immediately, or the control group, who received WGS after 2 years of conventional care.[5]

II. Interview Study

Qualitative interviews were conducted with a subset of families involved in the DDi arm of the SeqFirst project. 22 parents of children with developmental delays who received WGS results participated in semi-structured interviews, around 1 hour each. The interviews took place anywhere from 1 month to 33 months following the family's receipt of results from WGS.[11] The interviews covered topics such as parent's experiences with the testing process, understanding and processing of results, and how testing may have led to different types of support. These interviews were audio-recorded, transcribed, and de-identified before coding began. Of the total participants for these interviews, 11 received results that did not explain their child's developmental disability, 10 received results that did explain their child's developmental disability, and 1 received results that partially explained their child's developmental disability.[11]

III. Current Analysis

This thesis is a secondary qualitative content analysis of the 22 interview transcripts.[12] All data used in this analysis was de-identified. This analysis was guided by two frameworks, the medical and social models of disability,[6] both acted as the interpretive lens for parents' understanding of their child's disability. The analysis examined whether parents' responses revealed a medical model or a social model perspective. In this study, parents with medical model perspectives viewed their child's disability primarily as a biological issue needing to be treated. Social model perspectives emphasized acceptance, engagement with the disability community, and working to address societal barriers for their child. The post-WGS interviews were analyzed to see how these two perspectives appeared in parents' retrospective descriptions of their child as they discussed their family's experience before, during, and after sequencing.

IV. Coding Process

The data was managed and coded using ATLAS.ti Web (Version 9.26.0).The codebook was developed deductively using the two disability frameworks and three temporal periods: perspective before (before receiving testing/results), perspective after-immediate (right after receiving results), or perspective after-current (at the time of the interview). Each quote was coded twice, once for the model (medical or social) and once for time period. An inductive code,

“disability mentioned”, was added to pull out any explicit use of the word “disability.”

Coding was conducted by a single individual (the author) with prior qualitative methods coursework, but limited previous coding experience. The transcripts were not pre-read prior to coding. However, each transcript was read line-by-line and coded as they were read using the deductive codebook. The temporal codes (pre, post-immediate, post-current) were assigned based on questions asked and the time periods parents appeared to be describing in the text. Model codes (medical or social) were assigned based on the author’s interpretation of the framework definitions.

When quotes were ambiguous, the quote was coded with both models and reevaluated at a later point in the analysis.This was done by comparing quotes to those coded previously as well as regularly reviewing code definitions. Most feedback on the coding process was provided following the presentation of initial findings, focusing on the development of the three main result categories rather than specific individual coding decisions. As a result of this, the results section includes explicit explanations of why the representative quotes were coded as medical or social to support reading transparency.

V. Researcher Positionality

As a white American woman with graduate training in genetics and undergraduate training in disability studies, I bring a dual lens to this analysis. My

background includes work with the Deaf community and children with disabilities, which has shaped my commitment to disability-affirming research. However, I have limited experience working with parents who have children with disabilities or with people who have undergone genetic testing. Prior to coding, I held the assumption that parents would primarily seek genetic testing for diagnostic answers. To check this assumption against the data, I regularly reviewed code definitions and reviewed ambiguous quotes to ensure consistency throughout the coding process.

Table 1. Final Codebook

Code	**Code Definition**
Medical	Parent frames child's disability in the medical model of disability such as the use of WGS for diagnostic confirmation, a pathway to treatment, or understanding the child's disability in a medical capacity
Social	Parent frames child's disability in the social model of disability such as the use of WGS to advocate for accommodations, access services, or describe changes in societal/family attitudes towards disability
Perspective before	Parent describes period before receiving WGS results

Perspective after-immediate	Parent describes immediate reaction following receipt of WGS results
Perspective after-current	Parent describes current perspective at time of interview
Disability mentioned	Parent explicitly uses the word “disability” or “disabled”

Results

Three main patterns were identified which show differing dimensions of how parents view their child’s disability throughout the process of receiving WGS, all framed through the two models of disability analyzed.

I. Perceptions Prior to Results of WGS

The first pattern that was identified in this data had to do with the parents' perceptions of disability prior to the receiving of the results of WGS. Prior to the receipt of results, parents perceived their child’s disability through concern and medical uncertainty. This motivated them to pursue WGS as a path to a diagnosis.

One parent described their pre-testing experience as a search for answers also riddled with anxiety, affirming that: “...I was just trying to get answers for my daughter ...I just remember being super anxious...about it...if there was something going on with (Child)...” (Participant #13, Explained). This quote

illustrates that the period prior to WGS was full of both emotional distress but remained focused on pursuing diagnosis.

Another parent showcased somewhat contradictory feelings about the testing process, stating that: "...I was a little anxious at the possibility of finding something that maybe I wasn't prepared to know ...at the same time, knew that I would much rather know maybe than not know..." (Participant #23, Not Explained). This quote captures the complex emotions parents feel prior to receiving WGS results between wanting to get answers and being fearful of what the results might say.

II. Dominance of the Medical Model

The second pattern identified was the dominance of the medical model in parents' retrospective discussion of their child's disability throughout the WGS process. The majority of parents (16/22) consistently spoke about their child's disability in a medicalized way, aligning more with the medical model of disability. With this medicalized view, these parents more often described their child's disability as a biological problem needing treatment or diagnosis.

For example, one parent emphasized the value they found in WGS for identifying treatable conditions, explaining how they would recommend it to other families by stating: "[WGS] is a very good I think approach and for other families to go...to check if maybe there is other condition and...if not have teams this other condition that is affected your child, and then from there maybe other condition couple other way to support that kind of...condition, right? Maybe, maybe it's a

condition that the medicine can help…" (Participant #2, Not Explained). This quote shows how medical model framing focuses on clinical intervention being the main goal of genetic testing.

Another parent assumed that finding a genetic diagnosis would lead to a more clear and definite medical intervention by expressing that they: "…assumed that if there was a genetic reason…there might be treatments, or it might help inform the treatment a little bit more specifically…" (Participant #5, Not Explained). This parent's perspective towards treatment reflects a medical model framework because they seem to be prioritizing clinical outcomes over other ways to understand their child's disability.

III. Varying Trajectories for the Social Model

The final pattern identified varying trajectories for the social model (6/22), with some parents integrating more social model grounded thoughts immediately following receipt of results while for others it took some time for them to start using more social model types of beliefs toward their child. Of these 6 parents, 4 had received explanatory results from WGS. This suggests that clarifying results may play a role in creating space for social model thinking.

One parent reflected that receiving their results allowed them to shift away from focusing on medical causes by saying: "…we didn't have to worry about any sort of…medical stuff being…the cause for his issues, we have just focused more on…therapeutic approaches…and not really worrying too much about anything medical…" (Participant #3, Not Explained). This quote explores the idea that whole

genome sequencing reduced worry for some parents and allows for them to open up to more supportive and needs-based approaches for their child.

Another parent described how their perspective evolved following the receipt of their child's WGS results. They shared this by stating: "...And we're getting her the help she needs...we're seeing her make progress...We're getting her help and her therapists are coming over to help her out as well... all that has... brightened up the way we saw it at first..." (Participant #13, Explained). This illustrates a delayed integration of social model thinking because the parents only changed their thinking after seeing tangible improvements from therapeutic support. This parent attributes changes in their viewpoint to improvements from therapeutic approaches, so the value of WGS may be indirect here. However, this shift was likely related to WGS indirectly because they received an explanatory result, likely leading to greater accessibility to services.

Discussion

I. Summary of Findings

This analysis revealed three main findings. First, prior to the receipt of WGS results, all parents consistently framed their child's disability through concern and uncertainty. Second, most parents (16/22) remained consistently aligned with the medical model throughout their WGS process. Third, a smaller group (6/22) showed higher engagement with the social model within either the post-immediate or post-current period.

II. Connection to Literature

This study found most parents remained aligned with the medical model of disability over all time points discussed. The dominance of medical model framing aligns with research suggesting that genetic testing may reinforce medical views of disability.[8] In addition, research has found that parents who have received a genetic diagnosis for their child tend to focus more on things like treatment and medical management and less on acceptance and empowerment.[6] These findings suggest that for many families, WGS acts as a reinforcing factor for an existing medical model framework.

Parents who expressed perspectives aligned with the social model were most likely to do so immediately following the receipt of their results or at the time of the interview. This integration of social model thinking is consistent with research suggesting that the framing of disability may change over time.[13] In this study, however, social model alignment was still less common than medical model alignment (6/22). This finding expands upon the literature by presenting that while WGS often reinforces medical model tendencies, it can also open up some parents to social model thinking following the receipt of their child's results. This finding could be explored further in the future by looking at what factors may contribute to the variation in thinking for these particular individuals.

III. Limitations

There are several limitations to this project. The first and main one to be considered is that this is a secondary analysis of data that had already been collected. I was not involved with the creation of the interview guide or the interviews themselves. With this comes the fact that these interviews weren't specifically about disability, so some information formed into patterns may be based on subtext rather than direct testimony from the parents. Another limitation of this project is that there is only one coder for the data and because of this there was no way to assess inter-coder reliability. In relation to this, the distinction being made between medical and social model framing wasn't always clear. Many quotes involved elements of both models or fell somewhere in between which required in-real-time interpretive judgment. A different researcher may have coded some quotes differently, reflecting the subjectivity that is a part of qualitative research. A further limitation of this project is that these interviews were all performed following WGS, with questions being asked about the different parts of the process. Though the parents were asked about the different time points throughout the WGS process, they weren't collected in-real-time which introduces a factor of recall bias compared to if they had been interviewed at the different time points. The period of time from receiving results to participating in an interview was varied (1 month to 33 months). This wide range of times between results and interview means that the 'post-current' time point reflects different lengths of time to process results. A parent who received their child's

results 1 month ago compared to almost 3 years ago are likely to be in much different places with their child's health, which makes comparing them directly somewhat difficult. With the limitations of this project, there is a need for further research in this area with direct qualitative methods aligned with this sort of question specifically.

IV. Future Directions

Generally, future research in this realm should focus on longitudinal studies that would allow researchers to follow parents over time and see how their perspectives shift because of different factors, including things like receiving genetic testing. Ideally, a future study to answer the question asked in this thesis would look like a longitudinal study following parents over time, throughout the process of receiving whole genome sequencing or some other type of genetic testing. This study would be specifically designed to look at perspectives surrounding disability and could be done using a mixed-methods approach. This approach would allow for parental perspectives to be measured in a quantitative way through surveys as well as still including qualitative work through interviews just with a more defined "disability" based viewpoint.

Conclusions

I. Summary

This thesis looked at how parents of children with developmental disabilities perceive their child's disability following whole genome sequencing.

The findings of the analysis suggest that parents are much more likely to be reinforced in a medical model thinking surrounding their child's disability following the receipt of whole genome sequencing. If they do exhibit social model thoughts it is most common following the receipt of results, and prior to receiving their results parents typically viewed WGS as a path to treatment, not just understanding their child's disability. In the broader context, these findings suggest that medicalized views of disability remain common among the parents of this study.

II. Final Thoughts

As whole genome sequencing becomes more common in pediatric diagnostics, understanding how it affects parents' perceptions on disability is important. Ultimately, this study contributes to our understanding of how much work there is to be done within clinical spaces in order to show that a specific diagnosis isn't always the result needed in order to best support individuals with disabilities. Looking forward, researchers should expand work within the disability community, with inclusion of these individuals' perspectives, in order to open parents' minds to greater possibilities for their children with disabilities.

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Appendix A: Participant Characteristics (n=22)

	Category	DDI N	DDI %
Gender	Female	16	76
	Male	5	24
Age (years)	Under 24	0	0
	25-34	7	33
	35-44	10	48
	45-54	4	19
Race	Asian	1	5
	White	19	90
	Other	0	0
	Prefer not to reply	1	5
Ethnicity	Hispanic or Latino/a	3	14
	Non-Hispanic or Latino/a	16	76
	Prefer not to reply	2	10
Level of Education	High school graduate/GED	1	5
	Some college	9	43
	College graduate	5	24
	Post-graduate (e.g. MA, MS, PhD)	6	29
Annual Household Income	<$50,000	2	10

	$50,000-$100,000	4	19
	$100,000-$200,000	7	33
	>$200,000	7	33
	Missing	1	5
Explained Status	Explained/Partially explained	10	48
	Not explained	11	52