Understanding parent/caregiver support needs during genome sequencing in a pediatric research setting

Priyanka Murali

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Reading Committee:

Joon-Ho Yu, Chair

Gail Jarvik

Stephanie M. Fullerton

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Abstract

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Priyanka Murali

Chair of the Supervisory Committee:

Joon-Ho Yu

Department of Epidemiology

Pediatric patients benefit from genome sequencing (GS) for disease diagnosis, treatment guidance, and reducing diagnostic delays. However, parents and caregivers navigating this complex system face unique challenges, including informed consent, understanding results, and managing expectations. The variability in genetic service delivery in different clinical contexts can impact the parent/caregiver experience. Existing research often examines implementation factors like clinical utility, provider perspectives, and ethical/social concerns but tends to focus on specific aspects of genetic testing or lacks a comprehensive look at parental needs during GS specifically. This dissertation aims to address this gap by examining parent/caregiver needs throughout the entire pediatric GS process, from pre-test counseling to post-test follow-up as well as in different clinical settings. It integrates findings from a scoping review of existing literature examining parental needs during both pediatric whole exome and genome sequencing and qualitative interviews conducted within the SeqFirst project, which investigates GS as a first-line diagnostic tool in children with atypical development and infants admitted to the neonatal intensive care unit (NICU). My results identify key themes: parents need clear, empathetic communication and tailored information, emotional support, and logistical guidance at every stage of GS. Findings highlight the interconnected nature of informational, emotional, and logistical needs, underscoring the importance of addressing them comprehensively. Recognizing and responding to these needs can inform patient-centered implementation practices, ultimately improving healthcare experiences and outcomes. Addressing gaps in current support strategies, especially in the NICU and developmental disorder settings, is crucial as GS becomes a mainstay in pediatric care. This study advocates for tailored interventions to support parents, ensuring effective communication, emotional well-being, and navigational assistance through complex healthcare landscapes

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Chapter 1 – Background and Introduction

Pediatric patients routinely benefit from diagnostic genetic testing, including Exome and Genome Sequencing (GS), to identify diseases, guide treatment, end diagnostic odysseys, and reduce comorbidities.[1,2] During this process, parents and caregivers of pediatric patients must navigate a complex system of genetic services delivery, including interactions with a variety of key players. Since systems of genetic services delivery can differ in the way care is delivered, by whom care is delivered, and by patient entry point into the system,[3,4] parent/caregiver perspectives, experiences, and needs during the GS process can be complicated. The parent/caregiver experience is particularly important during the process of pediatric GS because of the potential complexity of the test result and the need for continuous long-term follow up by PCPs and other medical specialists (genetics providers among them).[5] In addition, parents/caregivers face many challenges, related to informed consent, return of results, secondary findings (variants discovered during testing that are of potential medical significance but unrelated to the primary reason for conducting the test), and unfulfilled expectations.[6] Researchers have studied various elements of implementing clinical genetic care, including assessing success factors like recruitment, enrollment, outcomes, and informed decision-making;[7,8] implementation challenges for clinical genetic testing (e.g. barriers and facilitators for participants and providers);[9] education and resources for various stakeholders around issues of informed consent,[10] understanding of results, data sharing, and clinical follow-up.[11,12] However, many of these studies are a) not specific to genome sequencing; b) limited to specific clinical indications; or c) an evaluation of general perspectives on GS rather than the process of GS itself.

Understanding parent/caregiver perceived needs is urgent because sequencing-based testing is being implemented now. The Pediatric Exome/Genome Sequencing Evidence-Based Guideline Work Group, comprised of members of the American College of Medical Genetics (ACMG) who specialized in clinical genetics, psychiatry, and neurology, used the Grading of Recommendations Assessment, Develop and Evaluation (GRADE) framework to develop and present evidence summaries and healthcare recommendations for clinical tests that improve patient outcomes and expand potential

treatments and stated that exome and genome sequencing should strongly be considered the first line of testing for patients with congenital anomalies, developmental differences, or intellectual disabilities.[13]

There are certain limitations in the literature currently. Existing studies examining implementation of clinical genetic testing in pediatrics have typically focused either on clinical utility and diagnostic yield[14,15] or psychosocial/ELSI concerns of patients.[16,17] Some studies examining patient experiences have tended to focus on specific indications or specialties,[18,19] while others have looked at experiences of health systems and experiences of patients (adult genetics) and parents/caregivers (pediatric genetics) as they navigate clinical genetic testing but have not focused on GS specifically.

Beyond this, there have been studies examining parental perspectives on GS for their children. These analyses have found that parents opt for their child to have GS given that they expect benefit for their children, but a lack of clear guidelines around informed consent and disclosure of secondary findings make it so that parental expectations are not always met,[20] indicating that these areas require further examination. Additionally, existing studies have examined a broad swath of concerns and challenges related to the implementation of clinical genetic testing. Although not all are specific to GS, the types of concerns that they examine are related, given the similarities in the nature of clinical genetic testing. However, these studies tend to focus on a single category of concern (clinical utility, diagnostic yield, ethical concerns, guideline development, for example) and rarely examine parent/caregiver perceptions and needs related to the process of pediatric GS specifically.

Implementation research about clinical genetics and genomics has not often been specific to GS or pediatrics, nor has it often contextualized GS in the broader delivery of healthcare. As many studies focus on one specific category of clinical indication, the recommendations made by the study are specific to those indications. Other studies focus on a single category of implementation concern, such as diagnostic yield and clinical utility, and the views of different stakeholders (i.e., patients, providers, insurers) or the Ethical, Legal, and Social Implications (ELSI) considerations and challenges that arise with GS. Additionally, although there are studies that examine parent or caregiver perspectives of GS, they have tended to focus on psychosocial needs, expectations, and understanding of GS testing.[21,22,20] While these contributions have led the field to the threshold of implementation, to date no studies have examined parent and caregiver perspectives and needs specific to the pediatric GS *process* (preceding,

during and following GS) rather than diagnostic yield/clinical utility/psychosocial and ethical concerns. Therefore, empirical research to inform specific process-related questions that consolidate these concerns under the specific context of pediatric GS is necessary to understand different categories of parent/caregiver needs and perspectives in order to hasten improvements to GS implementation.

Importantly, this study sits in a space that is informed by the above outlined concerns. It is focused on asking parents and caregivers about their perception of the GS process, interactions with stakeholders, and how navigating this process may or may not have met their expectations. Since this research has been conducted within the umbrella of SeqFirst project, a research study examining GS as a first line of genetic testing, we are able to examine perspectives in the context of a pediatric research setting and extrapolate these findings to broader clinical settings. An examination of parent/caregiver perceived needs has not been conducted in this type of liminal space. By capturing parent/caregiver perspectives, this study not only fills a significant research gap but also aims to develop actionable, patient-centered implementation strategies that can guide healthcare policies and improve outcomes in pediatric genomic care.

Chapter 2 – Aim 1: Parental/caregiver needs during pediatric genome-wide sequencing: a scoping literature review

INTRODUCTION

Advances in genetic and genomic testing have transformed the landscape of disease diagnosis, treatment, and prevention.[23] The development of these technologies has generated new opportunities for improved diagnosis of genetic disorders as well as target treatments in various clinical contexts.[24] Although genetic tests have long been ubiquitous in pediatric settings, these recent advances have changed the landscape of pediatric genetics. Technologies like whole-exome and whole-genome sequencing (together referred to here as genome-wide sequencing or GWS) are being increasingly used for pediatric patients with heterogenous medical presentations, given higher rates of diagnostic yield compared to previously available molecular and cytogenetic testing.[25] However, there are barriers that prevent parents and caregivers of pediatric patients from navigating medical systems as they go through the process of genetic testing.[9] These barriers and gaps are related to a lack of support at different points along the genetic testing process, including differential access, challenges to obtaining informed consent and returning results, as well as gaps in healthcare infrastructure.[7,8,26] As such, the clinical integration of GS requires a deeper understanding of how the implementation of genetic testing technologies is confronted and challenged by systemic problems that challenge adoption of new technologies by patients and their families.

GWS testing is often delineated into pre- and post-test periods which are thought to require different forms of genetic counseling.[27,28] The time period preceding testing (pre-test period) begins with an explanation of the method of testing used, the associated risks and benefits, and a discussion about the range of results that could be generated.[28] Then, a sample is collected and there is a period of waiting for results (interim period). In the post-test period, or the time after test results are available, information is shared and the families' understanding is assessed, expanded, refined, and/or corrected, potentially leading to the creation of a medical management and treatment plan.[28] The needs of parents of pediatric patients change as they go through this process, given that both the information and the context in which they receive it typically differ in the pre-test, interim, and post-test periods.[29]

Studies of parental perspectives and expectations in the context of single gene testing have identified various parental needs.[30,31] These include mitigation of anxiety and a need to better understand current and future impact of genetic testing results among parental needs during the genetic testing process. However, to our knowledge, no review of the current literature has examined the needs of parents and caregivers of pediatric patients as they have navigated the process of GWS testing across different clinical contexts. In addition, parental needs and experiences in different time periods of the GWS process have yet to be explored, including how these needs may be similar or different between time periods. Synthesizing this information may be useful in understanding how to improve parent experiences of GWS and at what time points during the GWS process to target potential interventions. We contend that as GWS becomes increasingly integrated into medical care, failing to understand patient-stakeholder perspectives may hinder effective implementation and exacerbate existing barriers to appropriate genetic and associated follow-up care.

Therefore, we conducted a scoping review of the peer-reviewed academic literature to examine the support needs of parents and caregivers of pediatric patients undergoing GWS. The primary aim of this review was to synthesize existing research to better understand these support needs throughout the GWS process. Additionally, we aimed to identify patterns or variations in these needs at different stages of the GWS journey. By doing so, we hoped to inform future research and clinical practices that can more effectively address the unique challenges faced by families navigating pediatric GWS.

METHODS

Scoping review question

The primary research question guiding this scoping review is: "What is the current state of knowledge about the needs and perspectives of parents and caregivers throughout the process of GWS in pediatric patients?"

To answer this question, we undertook a scoping review of the literature. A scoping review examines the extent, range, and nature, of a particular activity and works to identify gaps in the existing literature.[32] Scoping reviews are useful for examining emerging evidence when it is still unclear what other, more specific questions can be posed and addressed by a systematic literature review. Given that

the state of support needs in this specific context has not been fully examined, a scoping review can help to determine the depth and breadth of knowledge regarding this particular topic.

Search strategies

An initial search of electronic databases was conducted, following PRISMA search and screening guidelines for scoping reviews[33]. We generated search terms, built search strings, and conducted searches using these strings tailored to specific databases. PubMed, PsycINFO, CINAHL, Embase, and Web of Science databases were searched. Search terms were generated by first identifying concepts that encompass different parts of the question. These concepts included: “pediatrics,” “genome sequencing,” “parent or caregiver,” and “attitudes/needs/perspectives.” Relevant MeSH terms were identified based on these concepts of interest. Synonyms to MeSH terms were brainstormed and searched within each database. Some search strings were pre-built for specific databases. Other search strings were built by the first author and research librarian. Starting with MeSH terms, terms for other databases were modified to meet the specifications of the particular database. When terms from one database did not have an equivalent in other databases, those terms were included as free text in the search string. Specific search terms included:

> (Infant[Mesh] OR Child[Mesh] OR Adolescent[Mesh] OR Pediatrics[Mesh] OR Infan*[tiab] OR newborn*[tiab] OR "new-born*"[tiab] OR prematur*[tiab] OR preterm*[tiab] OR perinat*[tiab] OR neonat*[tiab] OR baby*[tiab] OR babies[tiab] OR toddler*[tiab] OR minors[tiab] OR minors*[tiab] OR boy[tiab] OR boys[tiab] OR boyhood[tiab] OR girl*[tiab] OR kid[tiab] OR kids[tiab] OR child*[tiab] OR schoolchild*[tiab] OR "school-age*"[tiab] OR adolescen*[tiab] OR juvenil*[tiab] OR youth*[tiab] OR teen*[tiab] OR "under-age*"[tiab] OR pubescen*[tiab] OR pediatric*[tiab] OR paediatric*[tiab] OR peadiatric*[tiab]) AND ("Genome sequencing"[MeSH Terms] OR "Exome sequencing"[MeSH Terms] OR "genome sequenc*" OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “Genetic services”[Mesh] OR "Genetic Counseling"[Mesh] OR “genetic services” OR “genetic counseling” OR “genetic counselling” OR "Genetic Testing"[Mesh] OR "Genetic Carrier Screening"[Mesh] OR “genetic testing” OR “genetic carrier screening” OR “genetic screening”) AND (“Parent child relations”[Mesh] OR Parent[Mesh] OR parent* OR mother* OR father* OR caregiver OR “care-giver*” OR guardian*) AND (“Health

knowledge, attitudes, practice"[Mesh] OR "Thinking"[Mesh] OR "Decision making"[Mesh] OR "Motivation"[Mesh] OR attitude* OR "needs" OR perspective* OR choice* OR experience* OR opinion* OR preference* OR concern* OR belief* OR desire* OR perception*)

For a list of complete search strings by database please see Appendix A. Titles and abstracts of all articles were searched.

Inclusion and exclusion criteria

We sought English-language studies focused on understanding the needs, perspectives, and attitudes of parents and caregivers of children who had undergone GWS, defined as either whole exome or genome sequencing. To be included, studies had to solicit the perspectives of parents or caregivers of pediatric (aged 0-18 years) patients who had undergone GWS. Studies that included multiple participant groups, such as pediatric patients themselves or clinicians or lay people in addition to parents/caregivers were included if the data for each group could be isolated. Studies must have returned results of GWS to parents/caregivers to be included. Qualitative, quantitative, and mixed methods studies were all included. Studies were excluded if they were opinion pieces.

Study selection

Database searches were conducted by the first author on in the following databases: PubMed, PsycINFO, CINAHL, Embase, and Web of Science on May 2nd, 2023 and search results imported into Zotero reference management platform (version 6.0.37). Duplicates were removed prior to uploading the titles and abstracts into Covidence, an online literature review management system. Titles and the abstracts of uploaded articles were screened for inclusion criteria and, if met, a full text review was conducted by the first author.

Data extraction

The following data were extracted from each included study using an extraction template: (a) bibliographic details (author, year, title, journal, DOI), (b) geographic location (c) study aim, (d) study design, (e) clinical subspecialty in which study was conducted, (f) participant relationship to child, (g) total number of participants in the study, (h) parent/caregiver experiences, expectations, and perceptions, (i) needs preceding, during, and following GWS (if explicitly stated), and (j) stakeholder interactions. The

extracted outcomes of this scoping review are parent/caregiver experiences, expectations, and perceptions, and parent/caregiver needs at different points during the process of genome sequencing.

Data about needs throughout the process of GWS testing was stratified based on whether this was a need preceding sample collection and genome-wide sequencing of the sample, during (in the interim between sample collection and return of results), or following return of GWS results. Extracted data were exported from Covidence into an Excel worksheet for analysis.

Data synthesis and analysis

Each article was reviewed three times to determine if all the data had been extracted and all relevant concepts captured. Excel was then used to calculate frequencies of different categories of extracted data, and descriptive and analytical themes.

Conventional content analysis

We conducted a conventional content analysis[34] of identified primary research studies to describe the state of knowledge of parents' needs throughout the process of pediatric GWS. As is typical with this approach, codes were developed inductively and sorted in categories based on how they were related and linked.

The extracted data from the needs preceding, during and following GWS categories were imported into Atlas.ti. 23.2.1 for Mac. Initial codes were developed based on a review of the data extracted from the first 10% of articles. The data were then coded line by line and more codes were added as necessary based on the content of the data. The data were then reviewed again to ensure that all relevant concepts had been captured. These codes were then sorted in appropriate descriptive categories and ultimately, broader themes. Specifically, analysis focused on code similarities and difference across all three different GS time points. These categories and themes were further refined and finalized following discussion between the qualitative research team.

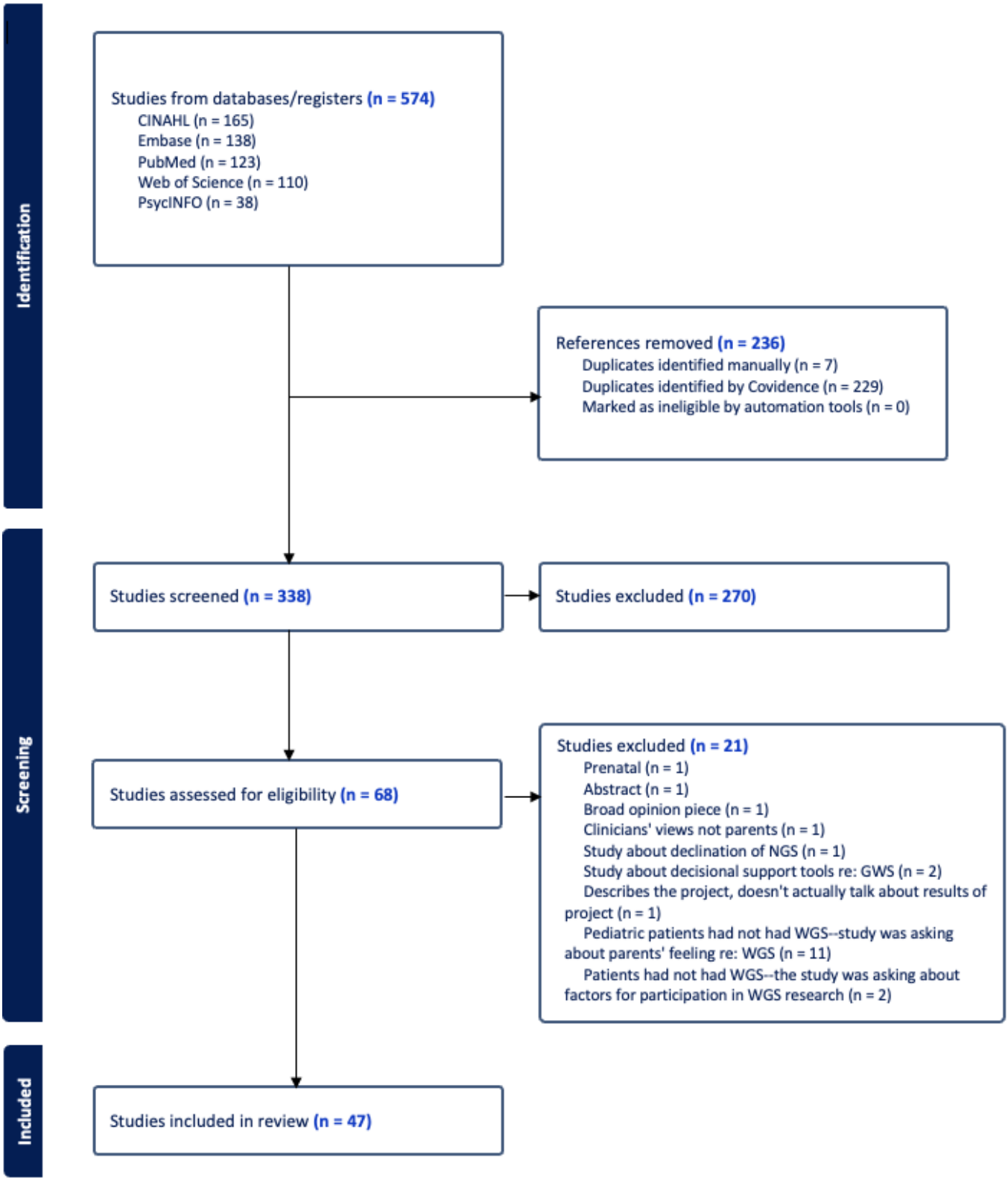
RESULTS

Search outcome

The database search returned 574 records. Once duplicates were removed, there were 338 studies remaining for title and abstract screening. Of those, 270 were deemed irrelevant based on the

inclusion and exclusion criteria, given that they were not discussing pediatric patients, did not conduct GWS, or were opinion pieces. This left 68 studies to be assessed for full text eligibility. A total of 47 were eligible for inclusion in the final scoping review. Figure 1 provides a flow diagram following the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines of the search and screening process.

Figure 1. Flow diagram following PRISMA guidelines demonstrating the results of the literature search.



Study characteristics

The 47 included studies provided data on the needs, experiences, expectations, and perspectives of parents/caregivers of pediatric patients who underwent GWS. Studies were conducted across five countries: the United States, Canada, the United Kingdom, the Netherlands, and Australia. Eight studies focused on GWS in NICU/neonatal settings, seven in oncological settings, six in rare disease settings, four in developmental settings, and four in neurological settings. Four studies involved GWS across multiple clinical settings, and the remaining seven studies were set in other medical settings, such as cardiology, in cohorts with congenital malformations, and cohorts undergoing newborn screening.

Twenty-one studies employed qualitative interviews, 15 utilized surveys, six used mixed methods, and the remainder used other approaches, including but not limited to focus groups. Table 1 provides a summary of article characteristics.

Table 1. Characteristics of articles

Country study was conducted in	N (%)
United States	25 (53)
Canada	10 (21)
UK	5 (11)
Netherlands	4 (9)
Australia	3 (6)
Clinical settings	
Other	14 (30)
Neonatal/NICU	8 (17)
Oncological	7 (15)
Rare disease	6 (13)
Neurological	4 (8)
Multiple settings	4 (8)
Developmental	4 (8)
Study design	
Interview – qualitative	21 (45)
Survey	15 (32)
Mixed methods	6 (13)
Other	5 (11)

Thematic analysis

Of the 47 articles, 37 mentioned parental needs related to the GWS process. Thematic analysis focused on these 37 articles to identify and classify needs that were preceding, during, and following GWS testing for pediatric patients. We coded more quotations about needs preceding (39%) and following (57%) in contrast to during GWS (3.6%). Identified themes centered on informational needs and emotional support, along with related subthemes.

Informational needs

Providers need to tailor communication and set expectations appropriately

An overview of these studies indicated that parents expected clear communication from providers to help them understand the overall GWS process. In the pre-test context, articles frequently emphasized parental reliance on providers to set and shape their expectations.[35,36,37] This included the need for a clear and easy-to-understand informed consent process to set expectations for testing[38,39,40] and a desire for clarity and acknowledgement that GWS findings might not lead to different treatment or provide different prognostic information.[41] Tailoring pretest counseling conversations was discussed in different articles, based both on patient understanding and according to specific clinical scenarios. For example, in NICU settings, parents requested succinct pretest conversations;[42] yet in the post-test context, studies reported overall parental dissatisfaction with the fact that they had received less information than desired.[43]

Parents need ways to gain information about logistics, prognosis, and next steps

In the pre-test context, some articles noted that a significant motivator for pursuing GWS[44] was the desire to better understand prognosis. In the post-test context, some articles found that parents also wanted information about logistics. Authors highlighted struggles with accessing medical services, medications, and dealing with insurance reimbursement issues.[35] Some studies highlighted a need for an intermediary to guide parents/caregivers through the healthcare system,[17] especially as the end of a diagnostic odyssey might mean the beginning of a new therapeutic journey requiring new care arrangements[45]. Beyond clinical care, parents sought therapies and services that often required a genetic diagnosis to gain access[46]. Some articles also reported a need for more information regarding secondary

findings[47] as well as a clear understanding of how results might impact the management of their child's condition, including planned follow-up, medical and otherwise.[45,48,49]

Guidance about future information changes is needed

Study authors discussed this topic exclusively in the post-test context. Study authors discussed parents' desires to understand how variant interpretations might evolve and how recommendations could change over time, as well as seeking clarity on how the relationship between the clinical genetics team and parents might continue in order to address these desires.[39] Some parents wanted explicit information on whom to contact and how to contact them,[17] while others preferred periodic outreach from genetics professionals.[45]

Emotional impact and support

Initial interactions with providers are key

In the pre-test context, initial interactions with the clinical team had an impact on parents' ability to process their feelings about the results in the post-test context.[50] Furthermore, interactions with providers influenced parents' perceptions of accessibility and coordination of care; when providers maintained regular contact, parents felt their care was more accessible.[40] In both the interim period while samples were being analyzed and in the post-test period, parents stated that improved accessibility of the clinic and associated providers improved rapport.[51]

Support from providers and peer groups is required

Some studies found a need for additional parental support in the pre-test context related to their anxiety about genetic testing[42] and a desire to alleviate feelings of overwhelm.[52] Forms of report were not specified. In the post-text context, articles mentioned a general need to feel emotionally supported by healthcare providers following return of results.[45,48] Other articles state that parents preferred that healthcare providers take a more active role in providing psychological support either by directing them towards resources or by making appropriate referrals.[53,54,55] Some articles mentioned the desire for connection with support groups for children with similar diagnoses.[44,51] Many studies also indicated that parents wished genetics providers would be the conduit for access to such support groups.[41,17,45,56] Other articles highlighted the importance of acknowledging parental frustration and discussing the possibility of uncertain outcomes in the pretest setting.[50] Discussions regarding return of results often co-occurred with

the concept of parental overwhelm and anxiety, typically in post-test settings. Parents stated that receiving a diagnosis did not eliminate negative emotions such as anxiety[36].

Notably, very few articles discussed needs in the interim period between sample collection and return of results.

Table 2. Themes, subthemes, and supporting quotes

Theme/finding name		Example quote from text
Informational Needs		
	Tailoring communication and setting expectations	*Keeping pre-test counseling brief and integrating this with a family systems approach can be very useful in a time-limited situation and to enable decision making for parents who experience decisional conflict or moderate-to-severe anxiety*[42]*.* *Families being approached for genomic sequencing may benefit from clinicians highlighting the increased probability of a rare or novel gene finding when compared to traditional targeted genetic testing*[41]*.*
	Information about logistics, prognosis, and next steps	*Parents across all interviews identified a desire to better understand their child's long-term prognosis as a key motivator to pursue whole-exome sequencing*[44]*.*
	Future information changes	*We also found that parents…may want more information on certain topics, such as which genes had been tested and whether reanalysis would be available to them in the future*[39]*.1/8/2025 9:22:00 PM*
Emotional impact and support		
	Initial interactions with providers	*…we feel that many of the inequities revealed in our study could be addressed within the current model of genetic counseling, for example with improved 'contracting,' or the dialogue exchanged at the start of a genetic session that establishes the patient-provider relationship*[52]*.*
	Support from providers & peer groups	*The importance of parent-to-parent support groups throughout the diagnostic journey was emphasized by parents in both cohorts, suggesting that peer connection may relieve social isolation for parents of children with rare disease*[41]

DISCUSSION

In this scoping review, we examined 47 articles to determine the current state of knowledge about parental and caregiver needs while their children as patients underwent GWS. We found that their needs were interconnected and evolved across different stages of the GWS process. Two overarching themes were identified: 1) Informational needs with the following subthemes: setting expectations and tailoring communication; information about prognosis, logistics, and next steps; and future information changes; 2) Emotional impact and support with subthemes: initial interactions with providers, and support from providers and peers.

This review highlights the fact that needs are varied across different stages but interconnected. In the pre-test setting, parents sought information to understand the types of potential results and the logistical aspects of the GWS process. Prior research suggests that effective communication strategies and realistic pretest counseling are essential for helping families navigate the uncertainty and potential complexity of the GWS process. This emphasizes the importance of communication and suggests a need for more overarching support from healthcare systems themselves as parents navigate the healthcare landscape.[57] In the post-test setting, their needs shifted toward understanding the specific results they received, their medical implications, and guidance on how to use or interpret these results in the future. These needs are consistent with those of other studies where parents sought guidance about whether the diagnosis should lead to changes in the daily management of their child and the changing nature of GWS research.[45] The review also suggests that the information provided and expectations set during the pre-test phase influenced how parents perceived and processed information in the post-test phase. This finding underscores the importance of tailoring communication to align with parents' understanding of genetic information and emotional state in the pre-test setting, as well as setting appropriate expectations, to better support comprehension and decision-making in the post-test phase. The extension of benefit from pre-test counseling into the post-test setting, specific to GWS, regardless of diagnostic yield has been seen in previously in the literature.[2]

In post-test settings, our findings suggest that parents also need appropriate expectation setting, now focused on both the immediate and long-term future. Parents expressed the need to understand how test results may impact medical care and access to non-medical services. They also sought clarity on

how GWS results and associated recommendations may evolve over time with future reanalysis or new data that could impact clinical guidance. Other articles examining post-diagnostic care in rare disease contexts have similar findings. They indicate that genetic conditions can pose a challenge to traditional care management models, given that patients often need follow up care and support from different categories of health professionals, likely from several different medical specialties, as well as by social worker providers, all of which requires a level of coordination challenging to organize in most health care systems.[58]

Our findings highlight that initial interactions with healthcare providers were also crucial for establishing rapport and offering emotional support to parents. Parents experienced anxiety and feelings of overwhelm throughout the GWS process. In the pre-test phase, this anxiety stemmed from the uncertainty of potential results, while in the post-test phase, feelings of overwhelm were linked to the complexity and volume of information provided. This suggests that anxiety persists across both stages of the GWS process and underscores the need for providers to deliver information empathetically to help manage parental anxiety and overwhelm. Furthermore, while parents expressed a desire for information, overburdening parents with information appeared to exacerbate feelings of overwhelm, indicating that a careful balance in communication is essential. Other similar studies report that balancing the amount of information provided does not result in lower parental understanding, and framing information in a way that allows patients to leverage their understanding can allow them to gain agency in their medical care.[59]

This review also emphasizes the importance of tailoring communication to match parents and caregivers' level of understanding and emotional state. This highlights the interconnected nature of informational and emotional needs, as meeting informational needs can directly influence emotional well-being. Parents especially valued and emphasized feeling cared for, having continuous relationships with clinicians, receiving empathetic communication, being kept informed while awaiting genetic test results, being connected with informational and psychosocial resources following results disclosure and follow-up.[60] This particular sentiment is echoed in other literature, where framing information in a way that enabled patients to leverage their understanding and gain agency over their medical decision-making also had psychosocial benefit.[59] Effectively addressing patients' informational needs helps them feel

supported throughout the process, and establishing good rapport early on further enhances their sense of emotional support.

Parents sought support not only from healthcare providers but also from peer support groups, suggesting that they benefit from perspectives beyond just the medical viewpoint. This points to the fact that parents gained support from a sense of solidarity through interactions with peers who experienced similar medical challenges. This is seen in other clinical genetics literature where parents recruited through peer support groups have emphasized the value of social and informational resources provided by these groups.[61,62] Notably, our review indicates that parents rely on healthcare providers to facilitate connections to resources outside the clinical setting, such as peer support networks. Thus, healthcare providers remain integral, either as direct providers of emotional support or as conduits to other supportive resources for parents.

Additionally, much of the literature appears to focus on needs in the pre-test and post-test phases. There are very few studies that have examined needs during the interim period between sample collection and return of GWS results, and as such, there is limited data on needs during this period.

Lastly, although several articles noted that parents required support at specific points during the GWS process, the specific nature of these needs was not articulated. This highlights a significant knowledge gap in the literature regarding parental needs in GWS. Addressing these identified gaps presents an opportunity for future research to further explore and define these specific support needs. Future studies may also benefit from directly engaging with parents of children who have undergone GWS, to gain insights into their experiences and assess how their needs align with or diverge from those outlined in this review.

LIMITATIONS

The findings of this scoping review should be considered in the light of several limitations. Included studies were conducted primarily in high-income countries which may limit the generalizability of these findings to diverse cultural and socioeconomic contexts. Studies with negative or inconclusive results, or those conducted in lower resourced settings may be underrepresented in the literature due to publication bias. This has the potential to skew the findings toward more well-supported themes,

potentially overlooking important challenges faced in less studied or less well-resourced environments. Despite these limitations, this scoping review highlights what is known in the literature and sheds light on what could be further explored with regard to parental needs in GWS.

CONCLUSION

In conclusion, this study identified informational and emotional support needs that spanned across different points of the GWS process in various clinical settings. The examination of these needs through contiguous points in the GWS process shed light on how informational and support needs evolve over the course of the process and how they are interconnected across time and through healthcare systems. By understanding these needs and how they arise, providers who are involved in different parts of the GWS process can better support families through the complex and often overwhelming process of pediatric genome-wide sequencing, ostensibly improving the implementation of GWS into pediatric care.

Chapter 3 – Aim 2: Parental needs during pediatric genome sequencing for developmental disorders: an interview study

INTRODUCTION

Developmental disorders describe a group of conditions that result in delayed cognitive, emotional, and even physical development in early childhood.[63] Although the etiology of these conditions varies, up to 80% are predicted to be related to an underlying genetic cause.[64] The emergence of genome sequencing (GS) and its use in genetics research over the last decade has substantially increased the diagnostic yield of genetic testing for children with developmental differences.[65] GS can confirm or establish diagnoses, lead to changes in medical management of the child, and anticipate associated comorbidities,[66] ultimately improving outcomes for patients and their families.[65]

The importance of partnering with parents of pediatric patients to shape and improve service delivery is increasingly recognized in healthcare.[67] An important part of implementing patient-centered care involves understanding parental needs, the feasibility of meeting them, and finally how they are being met.[60] As the clinical utility of GS is increasingly recognized by healthcare systems, implementation of GS for developmental clinical contexts requires understanding the needs of stakeholders, including patients and their families, to enhance the delivery of care.[60]

We previously conducted a scoping review to assess the current state of the literature regarding parental needs during the processes of both pediatric whole exome and genome sequencing, together termed genome-wide sequencing (GWS), in different clinical settings. Findings indicated that parents had specific informational and emotional support needs that were interconnected throughout the process of GWS. However, the specific nature of these needs and how they evolve during different phases of the sequencing process—pre-test, interim, and post-test—remain unclear. Nuances of these needs may be elucidated by talking directly with parents, whereby their firsthand experiences and insights can provide depth and context often not learned through secondary data alone. Therefore, we conducted a qualitative interview study examining the support needs of parents as they went through the process of pediatric GS for children with atypical development. By further exploring the current landscape of parental GS needs, specifically in the developmental disorder setting, this study identifies opportunities for provider interventions toward improving parents' healthcare experiences throughout the GS process.

METHODS

Study design and parent study

We conducted semi-structured interviews with parents of children with developmental differences who had received genome sequencing (GS) through the SeqFirst Study. The SeqFirst study offers GS as a first line test for eligible children with atypical development, termed the Developmental Difference or DDi cohort. Families were referred to SeqFirst from one of two Early Intervention Centers or from an academic neurodevelopmental clinic from April 2021 to November 2023. SeqFirst eligibility criteria included age <3 years and moderate or severe delay, or mild developmental delay plus abnormalities in growth, physical features, or organ systems. Exclusion criteria included prior evaluation or genetic testing for atypical developmental or a non-genetic explanation of patient characteristics.[68] GS was conducted on patients and, when available, parents as "trio" testing. Participants in the DDi cohort were randomized to either an experimental, in which they received GS immediately, or control group, in which receipt of GS was delayed. This portion of the SeqFirst study design was not relevant to this study on parental needs as questions about being in the control group were not analyzed.

Recruitment and data collection was conducted simultaneously with another qualitative study within SeqFirst examining recruitment experiences of participants, given the overlap in participants that would be contacted for recruitment and given the overlap in types of questions being asked in both studies.

This study was approved by the University of Washington Institutional Review Board.

Interview participant recruitment

We recruited parents of children with atypical development who had undergone GS through SeqFirst. A list of a subset of participants was generated in order to achieve a given proportion of families with explanatory, partially explanatory, or non-explanatory results. Each parent on this list was contacted by phone or by email using information provided by SeqFirst. All primary caregivers (mothers, fathers, and non-parent caregivers) of DDi children met inclusion criteria to be a participant in this study. We attempted to balance participant selection by aiming for an equal proportion of parents of children with explanatory and non-explanatory results. Participants provided verbal consent and received a $50 gift card after completing the interview.

Data collection

Semi-structured interviews were conducted to characterize parental needs as their children received GS. A short survey was used to collect basic demographic data about each participant. An interview guide (Appendix B) was developed based on *a priori* knowledge from Aim1 and tested with a six-person convenience sample including a research coordinator, one master's student, two doctoral students, and two SeqFirst co-investigators. The semi-structured interview guide included questions in the following domains: I. Warm-up (background information); II. Experience of SeqFirst recruitment and enrollment; III. Control group (for participants in the delayed control arm of the SeqFirst DDi cohort only); IV. Experience of sample collection and return of results; V. Impact of results on family and child's medical/non-medical care; and VI. Reflections on support and overall experience. Two study team members (K.M. and P.M.) conducted all interviews by phone between November 2023 and April 2024. Interviews were audio-recorded using a Bluetooth recorder.

This analysis included all data except for responses from Domain III, which focused on the experiences of parents who were randomized to receive GS in a delayed manner compared to those who received GS as a first line of testing upon enrolling in the study. This domain's primary relevance was to the parent SeqFirst Study. Interviews were conducted until participants' responses provided minimal to no additional information to answer the research question (i.e., data saturation).[69]

Data Analysis

Audio files were transcribed verbatim and transcripts were de-identified for analysis and reporting. One investigator (P.M.) read through all the transcripts, identifying codes based on interview questions as well as previous literature, including the Aim 1 scoping review. Three coding team members (P.M., B.C., and S.W.) coded the transcripts, with at least two researchers independently coding each transcript in Atlas.ti software (version 24.2.0). As coding progressed, additional thematic codes were added through iterative discussion with the research team. Coding differences were reconciled through discussion and, when coding consensus could not be achieved amongst coders, by involving the larger research team until consensus was achieved.

Coded quotations were output by pre-determined coding groups and emergent code relationships (e.g., co-occurrence). The present analysis presents themes characterizing parental needs in three

categories: informational, emotional, and logistical; across different time periods in the GS process: pre-test, interim, and post-test. This analytic perspective enabled us to examine the overlap of themes across previously identified need categories and their variation across pre-test, interim, and post-test periods. Findings were then refined through discussion with the research team.

RESULTS

Participants

We conducted 22 interviews with caregivers from 21 families. In two families, mother and father were interviewed together, while in one family, mother and father were interviewed separately. For the remaining families, only one parent participated in the interview. A total of 24 parents were interviewed. Demographic data were missing for three of 24 participants.

Full demographics are shown in Table 3.

Table 3. Participant characteristics (N = 21)

Participant type	N (%)
Gender	
Female	16 (76)
Male	5 (24)
Age (years)	
25-34	7 (33)
35-44	10 (48)
45-54	4 (19)
Race	
Asian	1 (5)
White	19 (90)
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)
Highest level of education completed	
High school graduate/GED	1 (5)
Some college	9 (43)
College graduate	5 (24)
Post-graduate (e.g. MA, MS, MD, PhD)	6 (29)
Annual household income	
<$50,000	2 (9.5)
$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)

Interviews elicited a range of responses, across the spectrum of emotional, information, and logistical needs. Notably, parent participants often relayed their needs through examples in which their needs were met, how they were met, and what they appreciated or valued during their GS experience (see Table 4. Summary of themes and subthemes.)

Emotional needs

Empathetic providers

Parents consistently appreciated when providers communicated empathetically, noting that emotional support was closely tied to how information was delivered. When they felt their provider communicated information effectively with both confidence and care, parents reported feeling more emotionally supported throughout the GS process.

> "…she kind of helped me understand how that was different from some of the other cases that I've seen on the Facebook group, and, you know, really kind of explained it in kind of layman's terms, so that I could understand what was actually going on. And I think that the whole appointment just gave me so much more hope. And I came out of that appointment feeling way more optimistic about our future." (*Qual_I01)*

Parents also reported that periodic check-ins from providers in the interim period helped alleviate feelings of anxiety.

> "I could imagine if people are maybe a little more hesitant or on the fence, like just touching, touching base and checking in. You know, 'I know, it's a long time to wait, we're looking forward to seeing you or whatever.' I think yeah, we were totally fine. You know, we're in it. But if people were on the fence, maybe that just more, more contact? Assurance. Say 'Yup, you know, we're still on schedule. We're still scheduled, just checking in to make sure everything's okay. Let us know if you have any questions.' Just things like that." (*Qual_I05)*

Connections to support groups

Parents of children with both explanatory and non-explanatory results commonly expressed a desire to connect with peer support groups or with other families with similar genetic or clinical diagnoses and to be a part of a community where they could provide help to other people in similar situations.

> "I'm on a Facebook group chat that has all the families with the condition that [NAME] as. And so when new people join, I'm able to read what their experience is like, or what they also went through and how they were able to get the genetic results. And so I've heard, like two families, I could relate to from that group. And I've also commented to try to help out some other moms that have questions…they [are] fairly new into the group chat, and they're dealing with their kiddos as well with the same stuff and so I've put my little info, what I know…" (*Qual_I13)*

Parents also voiced a specific interest in having a support group for families who had undergone GS through the SeqFirst study.

> "I think if there was a group…like in a group while going through the study, maybe? And maybe that would be something to like, look into because maybe if I had somebody that was going through the same thing I was going through maybe the diagnosis would be completely different, probably, but just having maybe that extra support of, what's happening with you, what's going on? Maybe that would have been better." (*Qual_I15)*

Informational guidance

Clarity in communication and expectation setting from providers

In the pre-test setting, parents expressed a desire for a clear understanding of why genetic testing was being recommended by healthcare providers, including how it might provide answers for their child. They also sought information about the details of the genetic testing process itself.

> "I think having a bit more of like, an understanding of why is the genetic screening? Like, what potentials could come out of it? I think that would have been helpful, because I still would have done it, but I think it would have prepared me a bit more." *(Qual_I01)*

Being "prepared" in this sense meant parents wanted more detailed information about the types of possible GS results: positive, negative, or uncertain.

In the post-test period, parents expressed a desire for a concrete understanding of how the information obtained from the testing would be practically applied to their child's medical care. Parents reported dissatisfaction when they perceived that their providers lacked sufficient awareness and information about the diagnosis to offer appropriate guidance. They also felt frustrated when the

information provided was vague or focused on the broad range of potential outcomes and genetic manifestations, rather than on actionable recommendations.

> "When we went for our genetic counseling, they're telling us all these very broad items that could be happening with [NAME]...if it was more narrowed down where they're talking about =John= more of a like, "Yes, we should be looking for this. Yes, we should be looking for that." Not, "Maybe we could be looking for this. I'm not sure." Like, there wasn't a lot of assurance during the counseling session…" *(Qual_I19)*

Written or online resources explaining GS testing or results

Parents indicated a desire for online resources or handouts on genome sequencing (GS) to which they could refer following the pre-test appointment or receiving test results. Some parents who were referred to online resources expressed appreciation for these materials because they could revisit the information if questions or uncertainties arose after their appointments with healthcare providers.

> "…I think it'd be helpful to have more information online, even just links to what really is genome sequencing? What are the limitations to this? And what are we looking at for timelines on this? And that maybe I was told, but…it's difficult to digest information, just being verbally told to me, when I have so much other stuff going on with, with my kid…so written material for me, especially online is really important for me to be able to retain the information, especially when it's information that I need throughout time…" *(Qual_I16)*

Understanding future use of genetic data

Parents expressed a desire to remain connected with the genetics team to stay informed about any changes in medical recommendations based on the existing genetic data.

> "When we met with Genetics team for second time, there were one or two more people with these genes studied, and they shared a couple more papers. And, but they basically...like there were no new information for us. Because like the results are very similar to what we already saw in previous studies. And yeah, this was helpful in the future, like somebody would find these new studies done for more people and to call us and say, 'Hey, we just...a new study was done and a thousand more people were found with this genes and these are new results.' Whether good news or bad news for us, it would be beneficial to have this communication." (*Qual_I20*)

Additionally, parents of children who received non-explanatory results often discussed the importance of staying in contact with the genetics team regarding the potential for future reanalysis of their child's genetic data.

Logistical needs

Clear logistical guidance

Parents expressed a need for clear logistical guidance and an understanding of next steps throughout the GS process. In the pre-test phase, parents wanted information about the expected turnaround time for results, how sample collection would be coordinated, and what to expect during the return of results appointment.

> "Yeah, that appointment…It was challenging for a couple of reasons for us. So, one, I think we didn't know how long that appointment would be. I thought it was just coming in to get our results. And it was much, much lengthier and probably I missed some instruction somewhere that explained what to expect. But I think that was just one thing that made it challenging." (*Qual_I08*)

In the post-test phase, parents of children with non-explanatory results wanted to know what conditions had been ruled out so that they eliminated the need to consult certain specialties. Those with explanatory results sought clear guidance on next steps, such as which medical specialists to see and which specific tests to pursue. As this parent explained:

> "I don't know what to expect any more than the doctors, because they said that there was only one other kid that [HOSPITAL] sees with the same diagnosis. And we were trying to get in touch with them, but they now don't want to. So, I'm like, I don't really know what to expect. I don't know how old this other kid was or is, you know, so I don't- I don't know what to expect. I don't know what other testing…I guess there was like, she needs an MRI…so she needs an MRI at some point. We got a scan of her heart and like a detailed ultrasound of her heart. So those are other testings, right? I just forgot." (*Qual_I15*)

Parents expressed interest in a care coordinator to help navigate the medical system after receiving GS results, even if results were negative. Parents stated logistical guidance was easier to understand when providers gave clear information about GS results. Clear logistical guidance also reduced anxiety and overwhelm in parents as explained by this participant:

“I was just very surprised to see how well it was organized and how everything was done for us, all the appointments and everything was set up, and everyone was following up. And I think it's just so important in any study, any trial, to make sure that the patient, or participant never feels lost, confused, what to do. And...we had resources available to us, if we had questions, everything was well explained, everything was prepared, everything was done for us, it's really just show up. So, we were just really grateful that it was so organized, so well organized.” (*Qual_I23*)

Table 4. Summary of themes and subthemes

Theme (need)	Subtheme (subneed)
Emotional support	*Empathetic providers*
	Connection to peer support groups
Informational guidance	*Clarity in communication & expectation setting re: different aspects of the genetic testing process*
	Written or online resources explaining GS testing or results
	Understanding future use of genetic data
Logistical guidance	*n/a*

DISCUSSION

Our findings indicate that informational guidance, emotional support, and logistical guidance are key needs throughout the GS process. Foundationally, parents desired empathic care and communication during the GS process to help manage anxiety and other emotions. Given that empathy and understanding from providers are key components of "good" care in healthcare broadly,[60] these parental desires lend credence to the idea that "good" care from providers during the GS process is similar to "good" care in general.[70] This cross-specialty overlap in the provision of "good" care allows all providers involved in the GS process to draw upon existing skills sets and familiar principles of pediatric care when trying to enhance patient care in the GS process,[60] despite GS-related appointments being arguably informationally and systemically complex.

Parents' desire for information about the practical implications of a diagnosis rather than a discussion about a range of potential clinical outcomes is mirrored in prior pediatric GS literature.[71] However, in many cases of developmental genetic diagnoses, prognostic uncertainty remains,[60] which limits the amount of information and reassurance GS can provide. This tension between potential outcomes versus practical implications suggests a potential mismatch between parental expectations of GS and its reality in the DDi setting. Setting clear expectations and exchange of information from providers at the start of the genetic testing process may mitigate this feeling in parents and clarify this discordance.[71] However, this parental desire may reflect an emotional need for control over their child's diagnosis rather than a purely informational need.[60,72] In non-genetics literature, emotional tools in conjunction with informational ones are proposed strategies to reduce diagnostic uncertainty.[73] Addressing this requires a patient- and family-centered approach, using empathy to educate parents about the diagnostic process and communicate potential uncertainties to better manage expectations and foster throughout the process.[73,74,75]

Parents' need for information about future impacts of GS results on their child's medical care highlights the importance of ongoing guidance from healthcare providers, especially given the potential for future variant reclassification and changes to care recommendations.[60] However, how best to address this need is a challenge given the practical burden of recontact for healthcare providers and the fact that guidelines and professional consensus surrounding providers' duty to recontact are nebulous at

best.[76,77,78,79] Given these challenges, it is difficult for providers to appropriately set expectations about future outreach. These challenges may be addressed through developing recontact guidelines that address patient concerns and mechanisms that facilitate provider-initiated recontact.[79] Additionally, these guidelines can be tailored to specific diagnoses, which may clarify allocation of responsibility among providers who recontact patients to ease their burden.[76]

Parents' emphasis on connection to peer support groups to alleviate feelings of loneliness and provide non-medical perspectives and insights on their child's diagnosis and future outcomes has been reflected in the literature[80]. In addition to needing peer support groups related to their child's specific diagnosis, parents also discussed the benefit of a support group for parents of children going through GS at large - regardless of clinical context - suggesting that GS itself may bring up feelings of isolation and need for peer perspectives similar to the way a specific genetic diagnosis might.[51] These findings underscore the importance of developing structured peer support programs that address both diagnosis-specific and GS-related experiences, ensuring parents have access to diverse and relevant forms of support throughout the diagnostic journey.

Parents' desire for a clear understanding of logistical next steps speaks to their need for smooth care transitions throughout the GS process. Given integration challenges within and across specialties, disjointed transitions of care have long been seen in genetics.[81,82] Potential solutions include clearer expectation setting and periodic check-ins from providers during different time points in the GS process to keep in contact with parents.[60] More broadly, as providers from many different specialties are involved in genetic testing, it is crucial for them to collaboratively address this need for integrated care.[70,82,83] At a health systems level, possible solutions include development of care plans that allow for providers from multiple specialties to be present at genetics appointments[70] or the inclusion of a care coordinator in genetics to make an inventory of parental needs and accordingly guide them through healthcare systems.[84]

Importantly, these findings suggest that parental needs are not independent of one another and that offering support in one category typically bolsters perceptions of support in another. Research has suggested that services focused on a specific clinical or social need may be insufficient when offered in isolation without a full understanding of the factors influencing individuals' health behaviors, self-

management capacity, and preferences.[85] Therefore, addressing parental support as a whole may be more effective than considering these needs independently, particularly given the interconnected nature of challenges faced by both patients and families as they navigate the medical system.[86] Addressing parental GS-related support needs have included examining who is responsible for these needs, which involves taking into account the individual roles of providers, health systems, and professional bodies in this ecosystem. However, conceptual frameworks describing the interconnectedness of actors within and beyond health systems highlight the importance of considering them together rather than in isolation.[86]

Given the opportunities and challenges to supporting families, future studies are needed. First, given the varied contexts in which GS is used, future research should focus on parental GS support requirements in non-DDI settings to determine areas of concordance and discordance with the findings of this study. Understanding similarities and differences in parental GS needs across different clinical settings can contribute to developing more robust frameworks and guidelines, enhancing their applicability and generalizability across different clinical scenarios. Additionally, toward developing cohesive and comprehensive strategies, future research should explore how different stakeholders, providers, health systems, and professional organizations interact and coordinate to meet parental needs related to GS. Designing solutions will require a greater breadth of knowledge about GS in different clinical contexts and will also require perspective from different stakeholders, ultimately yielding a greater breadth and depth of understanding about both the support needs and how to address them.

LIMITATIONS

All participants received GS through SeqFirst, which is a clinical research study. Receiving GS from a clinical research study introduced variations in how GS was offered to families. As such, their expressed support needs may not fully reflect those encountered in a standard clinical setting. Certain aspects of GS within a research context may have been emphasized more heavily in this study, while some elements relevant to GS in routine clinical care may have been underexplored. Additionally, study participants skewed toward white parents, which may have precluded the identification of needs more important to other less well-represented groups. Therefore, future efforts should prioritize the inclusion of

minoritized groups to ensure representation of diverse perspectives that may influence GS-related support needs.

CONCLUSIONS

This study is among the first to examine parental support needs during the process of GS for pediatric DDi patients. It provides a greater depth of insight into the support needs of parents of children with DDi as they go through the GS process. Mainstreaming of genomics into medical care presents an opportunity to understand the needs of key stakeholders and identify and develop interventions to address these needs. While further research across different clinical contexts and diverse clinical settings is needed, the findings of this study offer important evidence that can inform efforts to address current implementation challenges. Drawing on these principles of research and understanding them at a greater depth will go a long way in improving parents' care experiences in the GS and their health and well-being outcomes as well.

Chapter 4 – Aim 3: Parental needs during pediatric genome sequencing in the NICU: an interview study

INTRODUCTION

Genomic medicine represents a transformative approach to healthcare by leveraging genome sequence information from advanced tests such as genome sequencing (GS).[87] GS has the capacity to deliver molecular diagnoses that could not have typically been ascertained through traditional single-gene testing methods.[87] The neonatal intensive care unit (NICU) is particularly well-suited for the implementation of GS, given the higher prevalence of genetic disorders in newborns, the presence of genetic heterogeneity, and the fact that a complete clinical phenotype may not be present at birth.[88] GS in the NICU have been shown hold the potential to significantly reduce diagnosis times compared to conventional genetic testing, offer a broad diagnostic scope with a higher yield for identifying rare diseases, and inform clinical decision-making to support personalized treatment strategies.[89,65]

However, this diagnostic development raises questions about GS implementation strategies. Integration of GS into settings such as the NICU requires a thorough understanding of the specific context of clinical pathways and how stakeholders navigate them.[90] The needs of parent stakeholders are critical in the NICU,[91] and delivering patient- and family-centered care in pediatric settings necessitates understanding parental needs, assessing the feasibility of addressing those needs, and evaluating how effectively they are met.[67] With the growing recognition of GS's clinical utility, its implementation in NICU settings requires insight into parent stakeholder needs to enhance care delivery and improve patient outcomes.[60]

Different pediatric care settings are increasingly recognizing the potential for GS to enhance diagnostic capabilities and guide treatment strategies.[89,92,93] Diagnostic contexts and complexity of patients vary across different clinical settings within pediatric care,[94] which can influence the implementation and experience of GS in one clinical setting versus another. These contextual differences can influence how parents experience the GS process. Parental support needs also vary, shaped by factors such as the child's condition and the structure of care pathways.[60] This raises questions about how and whether meeting support needs in one context can be adapted to other pediatric environments.

Addressing these gaps is crucial to tailoring effective support systems and ensuring GS benefits all pediatric patients and their families.

Our previous work in Aims 1 and 2 highlighted that parental needs during the GS process are multifaceted, involving calls for informational, emotional, and logistical support. However, the specific nature of these needs and how they evolve during different phases of the sequencing process within the NICU—pre-test, interim, and post-test—remain unclear. Nuances of these needs may be elucidated by talking directly with parents, whereby their firsthand experiences and insights can provide depth and context often not learned through secondary data alone. Additionally, the specific nature of these needs and how meeting them might differ across different clinical contexts is similarly unknown.

Therefore, we conducted a qualitative interview study examining the support needs of parents of NICU infants as they went through the process of GS in order to understand their needs and to situate them relative to parental GS-related needs in other pediatric contexts. By further exploring the landscape of parental needs within the NICU setting, this study identifies opportunities for interventions toward improving patient and family healthcare throughout the GS process in pediatric care.

METHODS

Study design and parent study

We conducted semi-structured interviews with parents of infants admitted to the NICU who had received genome sequencing (GS) through the SeqFirst Study. The SeqFirst study offers GS as a first line test for eligible infants who have been admitted to the NICU, termed their NEO cohort. Exclusion criteria included age >6 months or clinical findings fully explained by physical trauma, infection, or complications of prematurity, and infants with a pre-existing PrGD via prenatal genetic testing or postnatal testing at their birth hospital were also excluded.[95] From January 2021 to February 2022, 407 infants were admitted to the NICU of a quaternary-level children's hospital, of which 235 met eligibility criteria. The study team met with 210 of these families, and 60% (126/210) were enrolled in the SeqFirst study.[96] GS was conducted on patients and, when available, parents as "trio" testing. Recruitment and data collection was conducted simultaneously with another qualitative study within SeqFirst examining

recruitment experiences of participants, given the overlap in participants that would be contacted for recruitment and given the overlap in types of questions being asked in both studies.

This study was approved by the University of Washington Institutional Review Board.

Interview participant recruitment

We recruited parents of the NEO cohort who had undergone GS through SeqFirst. A list of a subset of participants was generated in order to achieve a given proportion of families with explanatory, partially explanatory, or non-explanatory results. Each parent on this list was contacted by phone or by email using information provided by SeqFirst. All primary caregivers (mothers, fathers, and non-parent caregivers) of NEO children met inclusion criteria to be a participant in this study. Participants provided verbal consent and received a $50 gift card after completing the interview.

Data collection

Semi-structured interviews were conducted to characterize parental needs as their children received GS. A short demographic survey was used to collect basic demographic data about each participant. An interview guide (Appendix C) was developed by modifying the guide used in Aim 2, originally designed for parents of children with developmental differences who had undergone GS, to capture the experiences and perspectives of NEO parents. The guide was then piloted with a six-person convenience sample including a research coordinator, one master's student, two doctoral students, and two SeqFirst co-investigators. The semi-structured interview guide included questions in the following domains: I. Warm-up (background information); II. Experience of SeqFirst recruitment/introduction of GS; III. Return of results; IV. Impact of results on family and child's medical/non-medical care; V. Reflections on support and overall experience. Two study team members (K.M. and P.M.) conducted all interviews by phone between June and August of 2024. Interviews were audio-recorded using a Bluetooth recorder.

This analysis included all data. Interviews were conducted until participants' responses provided minimal to no additional information to answer the research question (i.e., data saturation).[69]

Data Analysis

Audio files were transcribed verbatim and transcripts were de-identified for analysis and reporting. One investigator (P.M.) read through all the transcripts, identifying structural codes based on interview

questions as well as previous literature, including the Aim 1 scoping review. We adapted the codebook, which was originally developed for Aim 2 involving interviews with parents of children in the Developmental Difference (DDi) arm of the SeqFirst study, to be applicable for transcripts in this study. Codes were added to capture the clinical experience of the NICU as well as the impact of genetic testing prior to the NICU. Certain other codes specific to the DDi cohort were not applicable.

Three coding team members (P.M., B.C., and S.W.) coded the transcripts, with at least two researchers independently coding each transcript in Atlas.ti software (version 24.2.0). As coding progressed, additional thematic codes were added through iterative discussion with the research team. Coding differences were reconciled through discussion sometimes involving the larger research team until consensus was achieved.

Coded quotations were output by pre-determined coding groups and emergent code relationships (e.g., co-occurrence). This analysis presents themes characterizing parental needs in three categories: informational, emotional, and logistical; across different time periods in the GS process: pre-test, interim, and post-test. This analytic perspective enabled us to examine the overlap of themes across previously identified need categories and their variation across pre-test, interim, and post-test periods. The themes and subthemes were then refined through discussion with the research team.

RESULTS

Participants

We conducted 18 interviews with caregivers from 18 families. In one family, mother and father were interviewed together. For the remaining families, only one parent participated in the interview, yielding a total of 19 parents interviewed. All interviews were included in the analysis. Participant characteristics are shown in Table 5.

Table 5. Participant characteristics (N = 19)

Participant type	N (%)
Gender	
Female	16 (84)
Male	3 (16)
Age	
Under 24	2 (11)
25-34	9 (47)
35-44	7 (37)
45-54	1 (5)
Race	
Asian	4 (21)
White	13 (68)
Other	2 (11)
Ethnicity	
Hispanic or Latino/a	2 (11)
Non-Hispanic or Latino/a	17 (89)
Highest level of education completed	
High school graduate/GED	1 (5)
Some college	11 (58)
College graduate	4 (21)
Post-graduate (e.g. MA, MS, MD, PhD)	3 (16)
Annual household income	
<$50,000	6 (32)
$50,000 – 100,000	5 (26)
$100,000 – 200,000	5 (26)
>$200,000	3 (16)
Explained status	

Explained/partially explained	16 (84)
Not explained	3 (16)

Interviews elicited a range of nuanced responses, across the spectrum of emotional, information, and logistical needs. Notably, SeqFirst-NEO parent participants often relayed their needs through examples in which their needs were met, how they were met, and what they appreciated or valued during their GS experience.

Emotional needs

Parents value empathy and time from their providers

Parents described the NICU as an inherently overwhelming experience, with the introduction of GS adding an additional layer of stress to their already heightened state of anxiety.

> "It was a lot. It's a lot to begin with. To just have a baby, to have a week-old, two-week-old newborn, to then add medical complications, being in the NICU, not being able to go home properly. And then to on top of that say, 'Okay, look here. There's this genetic diagnosis that kind of gives you a certain idea about what things might happen in the future.' You know, it's just one thing on top of another." (*Qual_I66*)

As such, they valued providers' empathetic care and appreciated when providers treated them as human beings.

Also, when providers took time to explain and re-explain information, parents felt more in control. As one participant explains:

> "I think, at the human level, we really thought our doctors cared for us. They really looked out. They would spend hours with us just explaining and re-explaining the same things. We had to hit these things many times to understand some of these things and to understand PA's [propionic acidemia's] extreme condition." *(Qual_I75)*

Parents want connection with support groups

Parents frequently expressed the need for connecting to peers to learn about raising a medically complex child, whether through online or in-person support groups or individual families.

> "And then there's bigger groups that I'm part of that just for short bowel syndrome and that one's active. So, like, you know, central line tips, TPN, things like that…and also, like, rare disease groups so, you know, even though our children don't have the same condition, just being a rare

disease parent and having a child with medical complexity…I feel like I've, I've found a lot of support both in person and online." *(Qual_I62)*

As the participant explains, support groups organized around syndromes or a shared experience of medical complexity served as an informational resource. They also found it emotionally helpful to connect with families whose children shared not only genetic similarities but also cognitive, physical, and emotional ones. As one parent stated:

> "…I could relate to other parents. Like I could relate to parents with the kids who have other severe syndromes…like I didn't really care if they had 6Q or not, but—it's the experiences talking and the expectations when they grow up. So that was way more useful than talking to, like, a counselor or something." *(Qual_I66)*

Parents want emotional support in intangible ways that they say clinicians cannot provide

Several parents reported relying on other family members for emotional support. Other parents expressed a desire for a sense of normalcy that providers could not fully offer. They sought normalcy not only in relation to their child's condition but also within the broader context of the NICU experience and the challenges of being postpartum so soon after birth. When asked if they wanted or needed anything while waiting for results, one parent explains:

> "Not anything that was providable. I mean, we were super, super anxious just waiting for the results. I felt like that part was very hard on us emotionally. The unknown was the most terrifying part, I feel like. But I mean, again, there's nothing that anybody can really do about that…" *(Qual_I54)*

Informational needs

Parents valued simplicity, transparency, and repetition from providers about the limits of their results.

Parents valued when genetics providers were transparent about what was known and unknown following GS results. This participant explained:

> "We wanted to know what our route was moving forward. And she was very upfront and transparent that like, 'This isn't everything. This might not be the exact case, but we're worried that this might be the scenario,' as we were finding things out. And so, we never felt like we were

left out to sea with any of the information or any of that. I mean, I really appreciated how transparent she was and immediate." *(Qual_I58)*

Parents felt supported when providers discussed gathering as much information as possible to direct clear next steps for care. Even when GS didn't yield a precise diagnosis, parents were reassured when results ruled out unneeded specialties. A parent described:

> "It was really nice to have the clarification of, when the stuff came back, like, 'Okay. There's no, like, chances for this,' or, 'There's no, results for any kind of condition.' And we were like, okay, well, that's a place to start." (*Qual_I47*)

Transparency helped parents feel more in control of their child's healthcare journey, for example when providers stated not know something and directed them to other providers for answers.

> "I feel like we had a lot of questions and just related to genetics and everything, we had all of our questions answered before and even after. Even if it wasn't the genetic counselor that was able to answer them, at least they told us like this isn't in our realm. You know, like this is more of probably like a clinical picture for the doctors to answer." (*Qual_I48*)

Several parents acknowledged difficulty in identifying their informational needs about GS. But parents appreciated having time to process new information and when providers were available to answer follow up questions. As this participant explains:

> "[Providers] would sit through the part where they kind of went over the paperwork and asked us all the questions. And then they would stay 15 minutes after that person left and make sure again that we felt comfortable going home and that we didn't have any other questions, that they didn't need to call them back and just clarify anything because sometimes in the moment you get flustered and then as soon as they walk out of the room, you're like, "Oh, shoot, I wish I would have went over this." (*Qual_I54*)

However, parents stated that in the case of children with rare diseases providing information about the diagnosis can be challenging for providers, given the limited baseline knowledge about these conditions themselves. Parents also discussed that these situations could bring parents both relief from the information provided by GS and anxiety when the information is limited or when their child's symptoms do not match existing literature.

> “I think it was partially a relief, knowing something. I think that relief also very quickly dwindled when we realized that not very many families had this or knew that they had it. And her symptoms still didn't line up perfectly with the results. So, there was still more questions on, does she have something else? They talked to us about how testing tissues can come up with different things than testing blood. I think it was a nice mix of relief and more anxiety.” (*Qual_I50*)

Parents want various tools to understand genetic information

Parents expressed their appreciation for having received resources like pamphlets or written materials that help them understand complex information.

> “I think they gave us enough information in the conversations that we had face-to-face, but also, like, all of the, you know, the pamphlets and the paperwork.” *(Qual_I48)*

Some parents also suggested that training or courses would help them become more familiar with genetics. As one parent notes:

> “I think if there was an opportunity even to attend some kind of seminar as a parent or training or orientation where if I can attend, I will. Even if it's a video that's sent to me so when I'm in my downtime and I can assimilate the information. ‘Cause I know there was other parents, you know, living at bedside and stuff and probably interacting with the genetics team. And there's some probably baseline information that everybody kind of learns and processes differently. And then it helps kind of prep us for any questions we might have and/or be able to apply that information to our child and maybe be more prepared to have, like…kind of an informed discussion about it.” *(Qual_I64)*

As described above, these resources would not only prepare parents to be informed about the GS but also better prepare them to navigate this journey.

Parents preferred sequential communication of information as compared to information dumping

Several parents described how information could be conveyed most effectively in the context of feeling overwhelmed by information about genetic testing. As this participant recalled:

> “[I]nitially, when they gave us all of our test results, it was so overwhelming. And they kind of said, ‘You know, although we're going all of—over this all right this second, it doesn't mean it's the last

time you're going to hear it, so don't expect to retain it all,' which I felt like was really helpful 'cause I was kind of like, you're in shock, you know?" *(Qual_I54)*

In addition to recognizing that parents could be overwhelmed and reassuring that information would be repeated, providing information as it became available, sequentially rather than all at once, was thought helpful by parents. As this parent described:

> "Oh, the first time was about [Child's NAME], and the second time was about [Parent's NAME]. And then a couple weeks later, they set up an appointment, a phone call to go over the preliminary results that came back. And so, they knew that there was an abnormality. The typical one on the 11th chromosome was this MEN2B one, which is this really bad scenario for either of them to have. And so that's what she told us when we were leaving the hospital. And then about two weeks later, when they had the full panel back, they were able to tell us, 'It's actually not this MEN2B abnormality. It's just a different one that we've never seen before, um, that has likely led to you guys having the kidneys missing and the Hirschsprung's disease.' So we had, like, three interactions with the same woman early on." (*Qual_I58*)

As noted in above (Emotional needs), parents also stated that it was helpful when providers would reiterate and re-explain information sometimes "hit[ting] these things many times" *(Qual_I75)* allowing it to become familiar.

<u>Logistical needs</u>

Parents desire a clear understanding of next steps

Parents consistently emphasized their appreciation for clarity about next steps, both in the NICU and after return of GS results. As this participant describes:

> "…the process has been very easy. Like, they, they run us through the information. They tell us, 'Okay, like, there—these are the steps, and this is what's going to happen. This is how long the test and the results will get back to you. Once we get the results, we'll give you—we'll go ahead and run through information with you as well and the results that we got.' So I just feel like everyone that has been through the process with us has been very informative and all the questions that I have are always answered." *(Qual_I52)*

Parents described that barriers to finding convenient times to meet providers in the NICU contributed to a lack of clarity about next steps and increased feelings of stress. Consistently, parents also stated that understanding next steps reduced their worries about the whole process. Additionally, parents felt reassurance that the process was on-going from regular check-ins during the interim period between sample collection and GS results. This parent recounted the benefits of such check-ins:

> "I feel like there was, um, somebody on staff that came and checked in with me at least weekly. I can't remember her name now but I feel like she was really easy for me to reach. I think I checked in with her periodically to see if the results are in or coordinating when I needed to do a sample for genetic testing. Having that person kind of readily available is helpful." (*Qual_I64*)

Parents desire a point-person to help them navigate the healthcare system

Parents without a designated point-person noted that having one would have been highly beneficial. As this parent explained:

> "There was a lot of questions that we had over the last couple of years where I would ask one of our providers, and they would say, 'Oh. You should ask your social worker that.' We never got a social worker, and I would tell them that. And they were like, 'Oh. That's weird. You should have gotten a social worker at the NICU, and you should still have one.' And we just never did, and we still haven't. And I don't know if that is a norm, if other families are getting that help, but having one person to refer back questions to that weren't always medical would have been really helpful like, having someone to help us with what different resources are out there for us…and all of the different things that we should have been signing up for, that we didn't know we were eligible for." (*Qual_I50*)

Parents also described the emotional toll of managing coordination themselves, anticipating that GS results would inform their child's care for the foreseeable future.

> "I think I felt like, 'Gosh, when is =John= gonna get a break?' Like, is life gonna be hard for him? Like, is this just the kind of life that he's gonna have? Because depending on what, like, what that looks like for him to be neuroatypical and have these needs, what does that mean for him? What does that mean for me as his mom?" (*Qual_I64*)

When available, parents consistently relied on complex care coordination teams, social workers or providers experienced in managing medically complex patients, to navigate the healthcare system. This support was to crucial to mitigate parents' sense of overwhelm from handling appointments and care processes.

> "And at times it was almost like too many resources. Like everybody has somebody to help, but at the same time, it's like to keep track of all of that and to call different people for different things we kind of use [HOSPITAL], the medically complex care team's coordinator as that central person to kind of help us coordinate everything else." *(Qual_I48)*

Further, parents described a range of providers who had acted as such including care coordinators, social workers, and even specialist physicians. As discussed by one parent:

> "You know, the communication of, like our immunologist, once we were done with the research study people and they had taken a sample, our immunologist then was the advocate to say, 'Okay, you know, I, I communicated with them, and this is where we're at in the process, and usually, it takes X amount of time.' And, you know, so that was— that was pretty much the only thing that I felt like was really super helpful and that we constantly kind of needed was just reassurance that we were moving forward 'cause it can feel like an eternity in the in-between." *(Qual_I54)*

Parents want teams from different specialties to work together

Given the medically complex nature of their children's diagnoses, parents frequently mentioned the need to work across various specialties. They emphasized the importance of easy coordination between specialties and appreciated collaborative efforts in developing treatment and care plans.

> "The teams all work well with each other too. For example, there was a medication [NAME] needed to take for her immunology a couple weeks ago, and she wasn't taking it. And everything online says, mix it with sugar, like chocolate, ice cream, whatever just get them to take it. And she can't have that because she has short gut syndrome. So, immunology worked with GI to kind of determine what would be the best plan of action for her to have this medicine, but also, you know, stick to her short gut diet. So, they're all very in the loop with each other and have a good treatment plan with each other." *(Qual_I62)*

While parents valued this coordination, they acknowledged systemic challenges that made even day-to-day coordination difficult, despite providers' best intentions.

> "Coordinated well is a challenge, though, because they're at the hospital, they will always be used with the doctors giving orders and being executed on the floor, right? That was always an issue. She needs an urgent medication. Doctors are saying, 'Give it ASAP, and it takes hour, hour and a half, to actually administer the administered that medication'. Those kind of challenges were a struggle. But I don't think that's on the on the providers themselves." *(Qual_I75)*

Discussion

The goal of this study was to deepen our understanding of parental support needs as their children, in neonatal intensive care, underwent GS. Empathetic and communicative care emerged as a fundamental need, one widely regarded as a cornerstone of patient-provider relationships in both genetic[60,5] and non-genetic healthcare settings.[97] This common regard is keenly relevant to the translation of GS into other pediatric specialties as many different providers may be involved in or touched by the GS process.[60] Furthermore, parents' GS-related needs appeared to depend on the variability of clinical contexts such as a child's medical complexity or the NICU environment.

The NICU's fast-paced environment can make it particularly challenging for parents to process complex genetic information in a specified time frame.[98] This challenge is likely compounded by the medical complexity and uncertainty surrounding rare or complex conditions, where baseline knowledge varies, and presentations differ across individuals.[99] Consistently, parents expressed a preference for receiving information sequentially in response to the large quantity of information they received (both genetic and non-genetic) in the NICU and follow up care. A two-tiered model for both informed consent and results disclosure could minimize the amount of information parents need process at one time thus potentially reducing information overload.[80,100]

While parents of children with a rare genetic condition almost universally seek out relevant peer support, study participants expressed a strong preference for connecting with families whose children share similar medical complexities, rather than those sharing a similar genetic diagnosis. This suggests that parents seek peer support that addresses the broader context of their child's care needs. Indeed, peer support in the NICU has been shown to empower parents, aid in medical decision-making, and

increase familiarity with community resources.[101] NICU providers ordering GS might consider directing parents to resources that offer guidance for their complex care needs as well as, when available, for their genetic diagnoses.

Additionally, parents emphasized the need for clarity and coordination to navigate the healthcare system. They appreciated receiving a clear understanding of next steps and care coordination (i.e., a point person) across medical specialties. The need for clarity and well-coordinated care underscores the importance of having a structured system with a delineated sequence of steps anticipated in their child's care and low-friction communication among providers. Study participants recognized the complexity of being in the NICU, with its own appointments and care coordination demands, that increase parental stress and logistical burdens[98,102,103] beyond those of GS alone. Although, integrating services across specialties can be challenging, both within and beyond genetics,[82] alleviating these burdens within genetics can include coordinated appointments involving multiple providers to collaboratively discuss a care plan during return-of-results sessions.[70] Scheduled multidisciplinary discussions in the NICU, where genetics can play a role, have demonstrated logistical benefits.[104] Use of a case manager may also help parents effectively communicate their needs and navigate complex care systems.[17] These approaches have been shown to enhance collaboration, improve continuity of care, and ultimately lead to better patient outcomes.

Parents' need for normalcy, one providers cannot meet, highlights the loss of a "typical" parenting experience in the NICU environment.[105] Parents' experience of losing "normal" parenting milestones has been shown to result in feelings of disempowerment[105] where parents feel a loss of control and stability. The question remains, what elements of care or support could help foster a sense of normalcy within the constraints of a medically complex setting. Although providers may not be able nor are expected by parents to meet their need for normalcy, research suggests that providers' support can help parents adapt their parenting vision to align with their current experience, in part, by acknowledging and validating the complexity of parents current situation and it's deviation from their expected experience.[106]

While NICU parents' support needs may, in part, depend on the intricacies of neonatal and complex care and DDi parents' needs may depend on their own unique contexts, when compared, parental support needs between NICU and DDi settings align more broadly. Parental needs—

encompassing emotional, informational, and logistical categories—were similar across all three aims of this dissertation, underscoring their importance across various clinical contexts. Although some of this alignment was expected, given that the relationships and shared methodology between aims, this consistency emphasizes the critical areas where parents seek support in the context of GS, regardless of their child's clinical setting. These findings suggest that while specific support mechanisms may need to be adapted to the nuances of each clinical setting, overarching themes such as the need for clear communication, peer connections, and comprehensive resource navigation remain vital. Addressing these parental needs can guide the development of targeted support strategies that are flexible enough to be applicable across different clinical environments, ultimately enhancing the overall effectiveness of care and parental experience during GS.

LIMITATIONS

All participants received GS through SeqFirst, which is a clinical research study. Receiving GS from a clinical research study introduced variations in how GS was offered to families. As such, their expressed support needs may not fully reflect those encountered in a standard clinical setting. Certain aspects of GS within a research context may have been emphasized more heavily in this study, while some elements relevant to GS in routine clinical care may have been underexplored. The participants of this study were skewed towards white parents, and largely female. This may have precluded the identification of themes that may be more important in other groups less well-represented in our interviews. Therefore, future efforts should prioritize the inclusion of minority groups, to ensure representation of diverse cultural perspectives that may influence GS-related support needs.

CONCLUSIONS

This study is among the first to examine parental support needs during the process of GS for infants admitted to the NICU. It uncovers a greater depth of knowledge about the support needs of parents of children who have been admitted to the NICU as they go through the GS process, providing specific insight into how underlying medical complexity impacts parental needs. This study also builds on previous works examining parental needs related to GS in the developmental difference setting. As GS

becomes more frequently used in clinical care, understanding these needs within and across different clinical settings offers opportunity to develop interventions to address them. While a further examination of different stakeholder perspectives (e.g., providers and administrators) is needed, this study offers important evidence that can inform efforts to address current implementation challenges. Gaining a deeper understanding of how to address these needs across clinical settings for parents will go a long way in improving their experience of pediatric care overall.

Chapter 5 – Conclusion & future directions

Our findings align with established frameworks of patient-centered care, which bolsters the credibility and applicability of these results. The Picker Institute's Eight Principles of Patient-Centered Care emphasize key elements—such as respect for patients' values, coordination and integration of care, communication, physical comfort, emotional support, family involvement, continuity, and accessibility[107]—that collectively underscore the importance of a holistic approach to meeting patient and family needs in healthcare settings. Similarly, the framework proposed by Santana et al. highlights the significance of effective patient-provider interactions, compassionate care, patient engagement in their own care, and coordination across specialties to improve care outcomes.[108] By demonstrating thematic alignment with these principles, our findings reinforce the need to address parental needs in a way that is supportive, responsive, and integrated, ultimately helping to ensure more patient- and family-centered approaches in the implementation of GS across different clinical settings.

Specific recommendations can be made to address the identified needs of parents engaging with pediatric genome-wide sequencing. First, there is a need for more robust guidelines related to the recontact of patients, regarding both explanatory and non-explanatory results. These guidelines should address the frequency and timing of recontact, as well as strategies for maintaining patient-provider relationships over time. Second, integrating care coordination into routine clinical practice could enhance the patient experience. While care coordination does not need to be led exclusively by genetics providers, it should be incorporated into care plans following the return of results. This approach would allow providers to evaluate the necessity and scope of care coordination and to ensure its integration into broader healthcare management. Finally, healthcare providers should prioritize empathetic care when interacting with patients and families. Although empathy cannot be distilled into a specific guideline and may appear self-evident, it serves as a critical foundation that influences how parents perceive and engage with other aspects of support.

While individual providers can support parents' informational, logistical, and emotional needs, a more systemic approach, as suggested by patient-centered care frameworks, may ease the burden of care on individual providers and enhance long-term support. Models where genetics are deployed as a

first line test (as it was in the SeqFirst study) often lack the ability to offer structured, continuous follow-up[109]—highlighted in our findings as critical to caregivers. Our research highlights that having a designated “point person” to coordinate care both within and beyond clinical settings can be beneficial. Existing care models for high-risk pediatric patients, such as those for pre-term infants or children with congenital conditions, may provide a template for addressing patient and familial needs in the case of infants with rare genetic disorders.[109,110] Such models incorporate structured, periodic assessments by a multidisciplinary team, offering consistent developmental monitoring and coordinated support.[110] This approach addresses multiple categories of needs by distributing responsibility across providers in a systematic and structured manner. Such models are promising, but further research is needed to develop and adapt such structural supports across varied clinical settings in GS.

Given the existing challenges and future opportunities to expand GS in various care settings, implementation studies are needed with an eye toward patient- and family-centered care. With greater awareness of parental needs in NICU and DDi settings, we have an opportunity to examine the perspectives of additional stakeholders in the care process (e.g., genetics and non-genetics providers, patient liaisons, and hospital administrators). At the individual level, exploring different categories of providers' views on parental needs and the resources required to address them could offer valuable insights into the feasibility of proposed solutions. As the primary facilitators of patient care, providers are uniquely positioned to assess both the benefits and challenges of implementing support strategies. Their dual role at the interface between healthcare organizations and patients enables providers to understand how system-level and logistical challenges impact the support available to families. Further research can explore how healthcare organization’s perceive families’ support needs to inform the development and testing of strategies that ensure needs are consistently met.

Finally, it is important to contextualize how specific these findings and thought processes are to the specialty of genetics. While GS is a test with broad applicability across multiple medical specialties, the needs and associated supports described by parents and reflected in the literature are not unique to genetics. These needs are comparable to those associated with other diagnostic tests ordered in various medical specialties that significantly influence future medical management. In this sense, GS represents one component of the broader healthcare continuum for patients and their families, and consequently, the

supports required throughout the GS process should be integrated into routine healthcare practices. While the novelty and increasing ubiquity of GS place genetics in a distinct position, as these technologies become more commonly used, there is a need to evolve our perception of them as routine rather than extraordinary tools within the healthcare landscape.

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Supplemental Information

Appendix A

Complete search strings by database

PubMed

(Infant[Mesh] OR Child[Mesh] OR Adolescent[Mesh] OR Pediatrics[Mesh] OR Infan*[tiab] OR newborn*[tiab] OR "new-born*"[tiab] OR prematur*[tiab] OR preterm*[tiab] OR perinat*[tiab] OR neonat*[tiab] OR baby*[tiab] OR babies[tiab] OR toddler*[tiab] OR minors[tiab] OR minors*[tiab] OR boy[tiab] OR boys[tiab] OR boyhood[tiab] OR girl*[tiab] OR kid[tiab] OR kids[tiab] OR child*[tiab] OR schoolchild*[tiab] OR "school-age*"[tiab] OR adolescen*[tiab] OR juvenil*[tiab] OR youth*[tiab] OR teen*[tiab] OR "under-age*"[tiab] OR pubescen*[tiab] OR pediatric*[tiab] OR paediatric*[tiab] OR peadiatric*[tiab])

AND

("Genome sequencing"[MeSH Terms] OR "Exome sequencing"[MeSH Terms] OR "genome sequenc*" OR “genomic sequenc*” OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “genetic screening”)

AND

("parent attitudes"[tiab:~2] OR "parent's attitudes"[tiab:~2] OR "parents' attitudes"[tiab:~2] OR "parental attitudes" OR

"parent perspective"[tiab:~2] OR "parent's perspective"[tiab:~2] OR "parents' perspective"[tiab:~2] OR "parental perspective"[tiab:~2] OR

"parent perspectives"[tiab:~2] OR "parent's perspectives"[tiab:~2] OR "parents' perspectives"[tiab:~2] OR "parental perspectives"[tiab:~2] OR

"parent needs"[tiab:~2] OR "parent's needs"[tiab:~2] OR "parents' needs"[tiab:~2] OR "parental needs"[tiab:~2] OR

"parent knowledge"[tiab:~2] OR "parent's knowledge"[tiab:~2] OR "parents' knowledge"[tiab:~2] OR "parental knowledge"[tiab:~2] OR

"parent experiences"[tiab:~2] OR "parent's experiences"[tiab:~2] OR "parents' experiences"[tiab:~2] OR "parental experiences"[tiab:~2] OR "parent choice"[tiab:~2] OR "parent's choice"[tiab:~2] OR "parents'

choice"[tiab:~2] OR "parental choice"[tiab:~2] OR "parent choices"[tiab:~2] OR "parent's choices"[tiab:~2] OR "parents' choices"[tiab:~2] OR "parental choices"[tiab:~2] OR "parent opinions "[tiab:~2] OR "parent's opinions "[tiab:~2] OR "parents' opinions "[tiab:~2] OR "parental opinions "[tiab:~2] OR "parent preference"[tiab:~2] OR "parent's preference"[tiab:~2] OR "parents' preference"[tiab:~2] OR "parental preference"[tiab:~2] OR "parent preferences"[tiab:~2] OR "parent's preferences"[tiab:~2] OR "parents' preferences"[tiab:~2] OR "parental preferences"[tiab:~2] OR "parent concerns "[tiab:~2] OR "parent's concerns"[tiab:~2] OR "parents' concern "[tiab:~2] OR "parental concerns"[tiab:~2] OR
“parent belief"[tiab:~2] OR "parent's belief"[tiab:~2] OR "parents' belief"[tiab:~2] OR "parental belief"[tiab:~2] OR
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"parent desire"[tiab:~2] OR "parent's desire"[tiab:~2] OR "parents' desire"[tiab:~2] OR "parental desire"[tiab:~2] OR
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"parent perception"[tiab:~2] OR "parent's perception"[tiab:~2] OR "parents' perception"[tiab:~2] OR "parental perception"[tiab:~2] OR
"parent perceptions"[tiab:~2] OR "parent's perceptions"[tiab:~2] OR "parents' perceptions"[tiab:~2] OR "parental perceptions"[tiab:~2] OR
(Parents[Mesh] AND (“Health knowledge, attitudes, practice”[Mesh] OR Attitude[Mesh])))

Web of Science

(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*)

AND

(“genome sequenc*” OR “genomic sequenc*” OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “genetic screening”)

AND

(parent* NEAR/1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*))

CINAHL (EBSCO)

((MH "Child+") OR (MH "Infant+") OR (MH "Adolescence+") OR (MH "Pediatrics+") OR TI(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*) OR AB(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*))

AND

((MH "Genetic Screening+") OR TI("genome sequenc*" OR "genomic sequenc*" OR "exome sequenc*" OR "genetic sequenc*" OR "gene panel sequenc*" OR "genetic screening") OR AB("genome sequenc*" OR "genomic sequenc*" OR "exome sequenc*" OR "genetic sequenc*" OR "gene panel sequenc*" OR "genetic screening"))

AND

(

(((MH "Attitude+") OR (MH "Health Knowledge")) AND (MH "Parents+")) OR TI(parent* N1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*)) OR AB(parent* N1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*))

)

Psycinfo (EBSCO)

(

AG(Childhood OR Adolescence)

OR

MA(“Infant” OR “Child” OR “Adolescent” OR “Pediatrics”)

OR

KW(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*)

OR

TI(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*)

OR

AB(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*)

)

AND

(

DE ("Genomic Sequencing")

OR

MA ("Genome sequencing" OR "Exome sequencing")

OR

KW(“genome sequenc*” OR “genomic sequenc*” OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “genetic screening”)

OR

TI(“genome sequenc*” OR “genomic sequenc*” OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “genetic screening”)

OR

AB(“genome sequenc*” OR “genomic sequenc*” OR "exome sequenc*" OR “genetic sequenc*" OR “gene panel sequenc*” OR “genetic screening”))

AND

(

((DE "Parents" OR DE "Adoptive Parents" OR DE "Expectant Parents" OR DE "Fathers" OR DE "Foster Parents" OR DE "Homosexual Parents" OR DE "Mothers" OR DE "Parental Characteristics" OR DE "Single Parents" OR DE "Stepparents" OR DE "Surrogate Parents (Humans)") AND (DE "Parental Attitudes" OR DE "Parental Expectations" OR DE "Health Attitudes" OR DE "Attitudes" OR DE "Health Behavior" OR DE "Health Knowledge"))

OR

MA ((Parents AND (“Health knowledge, attitudes, practice” OR Attitude))

OR

KW(parent* N1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*))

OR

TI(parent* N1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*))

OR

AB(parent* N1 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*))

)

Embase (Elsevier)

('adolescent'/exp OR 'child'/exp OR 'infant'/exp OR 'pediatrics'/exp

OR

(Infan* OR newborn* OR "new-born*" OR prematur* OR preterm* OR perinat* OR neonat* OR baby* OR babies OR toddler* OR minors OR minors* OR boy OR boys OR boyhood OR girl* OR kid OR kids OR child* OR schoolchild* OR "school-age*" OR adolescen* OR juvenil* OR youth* OR teen* OR "under-age*" OR pubescen* OR pediatric* OR paediatric* OR peadiatric*):TI,AB,KW)

AND

('whole exome sequencing'/exp OR 'genome sequencing'/de

OR

("genome sequenc*" OR "genomic sequenc*" OR "exome sequenc*" OR "genetic sequenc*" OR "gene panel sequenc*" OR "genetic screening"):TI,AB,KW)

AND

(('parent'/exp AND ('attitude to health'/exp OR 'attitude'/exp))

OR

(parent* NEAR/2 (attitudes OR perspective* OR needs OR knowledge OR experiences OR choice* OR opinions OR preference* OR belief* OR desire* OR perception*)):TI,AB,KW)

Appendix B

SeqFirst DDi

Parent Interview Guide

Modified: 1-18-2024

Interviewer/Moderator:

Record ID(s):
Date:
Start Time:
End Time:

BACKGROUND [DO NOT READ ALOUD]

Purpose: To understand the experience of recruitment/enrollment of families into the SeqFirst study DDi arm from the perspective of both patient's parents/guardians and EI/ESIT service providers. This guide focuses on the perspectives of parents/guardians.

* Indicates questions that should be prioritized if time is running short

Notes prior to interview:

Welcome & Introduction

Welcome and thank you for agreeing to participate in this interview.

My name is [*NAME*] and I work for [*INSTITUTION*].

Before we get started, I just want to review a few housekeeping details.

- I'm a member of the SeqFirst team, but I am a social scientist not an MD
- Our interview session will last 1 to 1½ hours.
- Your participation in this discussion is voluntary. You can refuse to answer any question, and you are free to leave/stop at any time.
- With your permission, we will record our conversation so we can transcribe it into a written version that my staff and I can review.
- Our discussion is confidential and no personal information about you will be associated with notes, recordings, or transcripts.
- If you're ready to start, I'll start recording.

[After start recording, confirm "I just want to confirm that you are allowing me to record this interview?"]

Questions

I'm going to start with a few questions about your child and how you got connected with the SeqFirst study. Then we'll zoom out a bit so I can learn more about how the SeqFirst experience fits into the broader picture of the care you have coordinated for your child

I. Warm-up

1. As a member of the SeqFirst study team, I know some information about [child] already, but it would be helpful if you could tell me about [child] in your own words. What is [child] like?

   Probes: Thinking back to the beginning of this journey…

   When did you first have concerns?

   How did you get connected to Boyer/CTC/Seattle Children's Neurodevelopmental Clinic? How is your child doing now?

II. Experience of SeqFirst recruitment and enrollment

Next I have a few questions about your experiences leading up to your decision to enroll in SeqFirst.

2. Before SeqFirst, had you ever thought about genetic testing for your child?

   Probes

   [If No] What was your impression of genetic testing back then? What was behind that, were there any specific experiences or information influencing your thinking?

   [If Yes, continue with] Was there something specific you were hoping to gain from genetic testing or genetic services more broadly?

3. My understanding is that you first heard about the SeqFirst study through your providers at [Boyer/CTC/NDV]. Is that correct? When did this conversation occur? What is your memory of that conversation?
   Probes:

   In that initial conversation, do you remember if they described GS to you?

   Did your provider talk about how the test results could impact you/your child? (e.g., potential changes to medical care)

4. When you were deciding whether or not to enroll your child in SeqFirst, what were some of the things you thought about or considered?
   Probes:
   What did you hope or expect from enrolling in SeqFirst?

   What were your concerns? Was there information you did NOT want to know (e.g. secondary findings)?

   Add a question here about the possibility of being randomized to delayed sequencing arm?

Besides the SeqFirst staff who walked you through the consent form, was there anyone else that you talked with about the project or whether or not you should enroll?

We're interested in getting feedback to make sure we are supporting families as much as possible when they are deciding whether or not to participate. Do you have any thoughts on things that could have been done differently that would have made the process better for you?
Probes: Amount/quality of information? Timing of conversation?

III. Control group (if applicable)

Next I'll ask some questions about your experience in the study after you decided to participate.

5. I see that you were assigned to wait for testing through SeqFirst. What has it been like to wait for testing?

   Probes: Have you sought or received testing elsewhere?

IV. Experience of sample collection and return of results (if applicable)

Next I'll ask some questions about your experience in the study after you decided to participate.

6. The standard procedure for sample collection is a blood draw. Did that go ok for you?

7. During the waiting period between collecting your samples and hearing about the test results, was there anything you wanted or felt like you needed during that time?

8. I see that you had an appointment with genetic counselor on X. What did you learn at this appointment?

   [Follow up] What did you do after receiving that information? How did you feel?

V. Impact of results on family and child's medical/non-medical care

9. [if not already addressed] Did you child's medical care change after receiving the result or diagnosis?

   [If yes] If so, in what way(s)? Were you surprised by any changes to care?

10. How has this information impacted you and your family?

Probes: Has it influenced any other decisions you've made for your child or about their healthcare? Decisions other family members have made? Family planning decisions? Insurance?

11. Have you shared this information with your child's non-medical providers/therapists?

    [If yes]: Do you think the diagnosis has impacted the kinds of things that [therapists] have been working on with your child?

12. Have you had any follow up with genetics?

13. *Optional:* One common reason why families want to get a specific diagnosis for their child is to get access to services. I'm curious if you've had any experiences in which having a specific diagnosis, or not having one, has made a difference in getting your child in to see specific therapists or get services?

    Probes [If child has clinical dx (e.g., autism)]: has the [clinical dx] vs. the [genetic dx] been more/less helpful in this regard?

VI. Reflections on support and overall experience

In this final section I'll ask you do some overall reflection on the experience of getting GS through SeqFirst.

14. *Would you recommend SeqFirst/GS to your past self, or other families like yours? How might you describe the advantages/disadvantages?

15. *Is there any information you wish you had known ahead of time that might have influenced your decision about whether or not to go down this path? Or that would have been helpful for making the journey smoother? Any advice you'd give to other families?

    [optional probes if needed:]

    After you received your GS test results, was there additional help or support you were looking for from the SeqFirst or genetics team?

    [Follow up] Did you get that support? If yes, from whom (e.g., Seqfirst study team, genetics, other providers)?

    [If got a diagnosis, ask] Have you connected with any peer groups?
    [If YES] How did you get connected to them?
    [If YES or NO] Did you get/wish you had had any support getting connected?

16. As we have had this conversation, have any new thoughts come up for you, things that you may not have realized or considered before we talked?

VII. Provider recruitment [ask wherever fits best in the interview]

17. We are also recruiting providers/therapists who work with children in SeqFirst to share their perspectives on how the genetic testing has influenced their treatment planning. Would you be open to us speaking with your child's therapists about this?

    IF YES -> send link to demographics and HIPAA release

    IF NO -> collect demographics over the phone

That is all the questions I have planned. Do you have any questions for me?

What is your age? ___	How do you describe your race? Select all that apply. • American Indian/Alaska Native • Asian • Black or African American • Native Hawaiian or Other Pacific Islander • White • Other • Prefer not to answer
What is your gender identity? • Female • Male • Non-binary • Transgender • Other • Prefer not to answer	What is the highest level of education that you have completed? • Did not complete high school • High school graduate/GED • Some college • College graduate • Post-graduate (e.g., M.A., M.S., MD., PhD)
How do you describe your ethnicity? • Hispanic or Latino/a • Not Hispanic or Latino/a • Prefer not to answer	What is your annual household income? • < $50,0000 • $50,000 - 100,000 • $100,000 - 200,000 • >$200,000

Closing and payment

- In the next 24-48 hours we will send $50 electronic gift cards to your email address.

Notes following interview:

Appendix C

SeqFirst NEO
Family Interview Guide
Modified: 6-18-2024

Interviewer:
Qual ID:
NEO ID:
Date:
Start Time:
End Time:

BACKGROUND [DO NOT READ ALOUD]

Purpose: To understand the experience of recruitment/enrollment of families into the SeqFirst study NEO arm from the perspective of both patient's parents/guardians and NICU service

providers. This guide focuses on perspectives of parents/guardians.

* indicates questions that should be prioritized if time is running short

Notes prior to interview:

Welcome & Introduction

My name is [*NAME*] and I work for [*INSTITUTION*].
Before we get started, I just want to review a few housekeeping details.

- I'm a member of the SeqFirst team, but I am a social scientist not an MD
- Our interview session will last 1 to 1½ hours.
- Your participation in this discussion is voluntary. You can refuse to answer any question, and you are free to leave/stop at any time.
- With your permission, we will record our conversation so we can transcribe it into a written version that my staff and I can review.
- Our discussion is confidential and no personal information about you will be associated with notes, recordings, or transcripts.
- If you're ready to start, I'll start recording.

[After start recording, confirm "I just want to confirm that you are allowing me to record this interview?"]

Questions

I. Warm-up

I'm going to start with a few questions about [child], your NICU experience, and how you were first introduced to the SeqFirst study. Then we'll zoom out a bit so I can learn more about how the SeqFirst experience fits in to the broader picture of care you've coordinated for your child.

1. As a member of the SeqFirst study team, I know some information about your [child], but it would be helpful if you could tell me about it in your own words. What is [child] like?

   Thinking back to your NICU experience, could you share (to whatever extent you feel comfortable) an overview of your family's experience at the Seattle Children's NICU?

   Probes (not to directly ask, but to get a sense): What was your duration of stay? What medical challenges did you encounter?

II. Experience of SeqFirst recruitment/introduction of GS

Next, I have a few questions about recruitment into SeqFirst. I recognize you had a lot going on at that time, so may not remember in a lot of detail, but I'll go ahead and ask you about it anyway. *(Note: If they don't remember, don't push for answer—this results in bad data)*

2. How do you remember hearing about the SeqFirst study?

   Probes: Who approached you?
   What is your memory of the conversation?

3. When during your NICU stay did that conversation occur? How was that timing for you?

   Probes: Would you have preferred an earlier or later discussion? Or would you say it was appropriate timing?

4. Do you remember how they described the GS to you? Did they talk about how it might impact [child]'s care?

5. How did you make the decision to enroll?

   Probes: What were some of the things you thought about or considered? What impression, if any, did you have of genetic testing before your decision to enroll?

6. What did you hope to gain from GS/genetic testing or getting connected with genetics?

   Probes: What were your hopes/expectations for genetic testing? What were your concerns?

7. We're interested in getting feedback to make sure we are supporting families as much as possible when they are deciding whether or not to participate in SeqFirst. What, if anything, could have been done differently that would have made the process better for you?

   Probes: Amount/quality of information? Timing of conversation?

III. Return of results

8. While you were waiting for your GS results from SeqFirst, was there anything you wanted or felt like you needed during that time? *(Note: keep in mind that TAT is 7-10ish days)*

9. How did you get your results?

   Probes: Did you have a follow-up conversation with anyone about the GS results? With whom (GC, NICU doc?)? What did you learn?

10. What was it like for you to get this information?

    Probes: How did you feel [emotionally]?

What kind of help or support did you receive from your providers? Was there additional help or support that you were looking for? Did you get that support? If yes, from whom?

IV. Impact of results on family and child's medical/non-medical care

11. How has your child's healthcare/medical care been since then?

    Probes: (it not already addressed) How is your child doing now?

    Probes: (For families with explained results) How did the results/receiving the diagnosis impact your child's medical care?

    Has this information had any impact on other therapies your child has received? For example, early intervention, physical/speech/occupational therapy?

12. What, if any, impact has this information had for you and your family?

    Probes: Has it influenced any other decisions you've made for your child or about their healthcare? Decisions other family members have made? Family planning decisions? Insurance?

    Are you currently under the care of a geneticist or genetic counselor?

    Probe: (if no) are there any providers who specialize in your child's diagnosis? Or who provide diagnosis-specific recommendations?

13. [If they got a diagnosis] Have you connected with any peer groups? How did you get connected? Did you get/which you had had any support getting connected?

V. Reflections on support and overall experience

In this final section, I'll ask you some overall reflection on the experience of getting GS through SeqFirst

14. Thinking back to where you were prior to starting this process, would you recommend participation in SeqFirst or GS to your past self? Why?

    Probes: Would you recommend it to NICU families like yours?

15. What information do you wish you had known ahead of time that might have influenced your decision about going down this path or made this process smoother for you?

    Probes: Would anything else have been helpful toward improving your experience of GS?

16. As we have had this conversation, have any new thoughts come up for you, things that you may not have realized or considered before we talked?

17. We are also recruiting providers/therapists who work with children in SeqFirst to share their perspectives on how the genetic testing has influenced their treatment planning. Would you be open to us speaking with your child's therapists about this?

    IF YES -> send link to HIPAA release, collect demographics over the phone

    IF NO -> collect demographics over the phone

That is all the questions I have planned. Do you have any questions for me?

What is your age? ___	How do you describe your race? Select all that apply. ▪ American Indian/Alaska Native ▪ Asian ▪ Black or African American ▪ Native Hawaiian or Other Pacific Islander ▪ White ▪ Other ▪ Prefer not to answer
What is your gender identity? ▪ Female ▪ Male ▪ Non-binary ▪ Transgender ▪ Other ▪ Prefer not to answer	What is the highest level of education that you have completed? ▪ Did not complete high school ▪ High school graduate/GED ▪ Some college ▪ College graduate ▪ Post-graduate (e.g., M.A., M.S., MD., PhD)
How do you describe your ethnicity? ▪ Hispanic or Latino/a ▪ Not Hispanic or Latino/a ▪ Prefer not to answer	What is your annual household income? ▪ < $50,0000 ▪ $50,000 - 100,000 ▪ $100,000 - 200,000 ▪ >$200,000

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