

“Is that something that should concern me?”: a qualitative exploration of parent understanding of their child’s genomic test results

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Summary

Genetic counselors are trained to deliver complicated genomic test results to parents of pediatric patients. However, there is limited knowledge on how parents perceive this information and what they understand about the results. This research aims to qualitatively explore parents’ experiences receiving genomic test results for their children. As part of formative research for the NYCKidSeq Study, we recruited a purposive sample of parents of 22 children stratified by child race/ethnicity and test result classification (positive, uncertain, or negative) and conducted in-depth interviews using a semi-structured guide. Analysis was conducted using grounded theory’s constant comparative method across cases and themes. Parents described different elements of understanding: genetics knowledge; significance and meaning of positive, uncertain, or negative results; and implications for the health of their child and family. Parents reported challenges understanding technical details and significance of their child’s results but gladly allowed their providers to be custodians of this information. However, of the different elements of understanding described, parents cared most deeply about being able to understand implications for their child’s and family’s health. These findings suggest that a counseling approach that primarily addresses parents’ desire to understand how to best care for their child and family may be more appropriate than an information-heavy approach focused on technical details. Further research is warranted to confirm these findings in larger parent cohorts and to explore ways genetic counseling can support parents’ preferences without sacrificing important components of parent understanding and overall satisfaction with their experiences with genomic medicine.

Introduction

Genetic counselors (GCs) undergo extensive training to educate patients about genetic and genomic concepts.¹ When GCs deliver test results to their patients, they aim to deliver the information comprehensively, guided by the notion that a solid knowledge base will facilitate patients’ informed decision-making about testing and treatment options. Improving patient understanding, therefore, has been a key goal of GCs. Unfortunately, there is a lack of consensus in the literature about what constitutes patient understanding of genetic and genomic test results. In the context of pediatric genetics especially, little is known about the scope of what parents need or want to understand about their child’s test results or the relevance to their lives.

Some quantitative research has instead measured cognitive control, a similar concept, which includes decision-making, planning, and contextualization of information.^{2,3} Others have focused on genomic health literacy, defined inclusively as “the capacity to obtain, process, un-

derstand, and use genomic information for health-related decision-making,” aiming to leverage genomic knowledge as a pathway to health promotion.^{4–6} Typically, however, studies of patient understanding in genetics focus more narrowly on knowledge of general genetic concepts,^{7,8} including genes and heredity,^{9–11} genomics,^{8,12,13} and genetic specialties such as cancer genetics.^{14,15} While it is broadly accepted that knowledge is connected to understanding, there is little consensus about other elements that constitute the larger construct of patient understanding, including important distinctions between perceived understanding, accurate interpretation, and meaning-making of results.^{16–19} The field of genomic medicine is expanding to new clinical areas and populations; thus, gaining clarity about the construct of understanding, particularly about genomic test results, is paramount.

Qualitative methods are particularly appropriate for exploring complex patient experiences with genomic testing and how people make sense of their results. Previous qualitative research in genetics has explored patient-reported outcomes such as confidence in understanding

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results, awareness of risks, and understanding of implications for family members.^{20,21} Some have investigated the ways cultural, linguistic, and racial or ethnic differences in understanding widen disparities for patients.^{6,22}

Regardless of methodology, even less is known about parent experiences with or understanding of their child's genomic test results.^{22,23} The limited research conducted with parents in pediatric settings has described parental motivations for and perceptions of genomic testing²⁴ and has highlighted specific challenges that remain unresolved, including parents' desires for additional post-test follow-up,²⁵ feelings of relief and worry evoked by genomic testing,²⁶ fluctuating emotions throughout the testing process,²⁷ and coping with guilt for believing they contributed to disease in their child.²⁶ Further, because most genetics research has been conducted in White, European-ancestry populations, the experiences of racial and ethnic minority parents have been largely unexplored.^{5,28} A variety of evidence shows that racial and ethnic minority parents' experiences in and out of the healthcare setting affect child health, including poor healthcare utilization among Black children when parents experience discrimination,²⁹ lower health insurance enrollment among Latinx children due to immigrant parent deportation fears,³⁰ and non-adherence to asthma medication among Hispanic children explained by their mothers' mistrust of the medical system.³¹

As the field of genomic medicine expands, genetic counseling practices will need to evolve responsively to align with the needs and goals of racial and ethnic minority families. To be successful, a rich evidence base is needed that describes diverse families' experiences. As part of the NYCKidSeq study, which is one of six clinical sites funded as part of the Clinical Sequencing Evidence-Generating Research (CSER) consortium,³² we interviewed a racially and ethnically diverse group of parents whose children had undergone genomic testing for a suspected genetic disorder. The aim of this research was to explore parents' experience of their child's genomic testing, with a focus on parents' understanding of results.

Subjects and methods

We conducted in-depth qualitative interviews with a diverse sample of parents to explore how they interpreted their child's genomic test results. This study was part of formative research to develop a novel web-based communication tool tested in a randomized control trial: the NYCKidSeq study.³³ The study was approved by the Institutional Review Boards (IRBs) of the Albert Einstein College of Medicine/Montefiore Medical Center (IRB 2018-8950) and the Icahn School of Medicine of Mount Sinai (IRB 18-00333).

Sample

We recruited and screened parents of children from physicians' patient rosters at two New York City (NYC) hospital systems: Montefiore Medical Center (Bronx) and the Mount Sinai Health System

(Manhattan). To be eligible, their child age 0–18 years needed to receive whole-exome sequencing, targeted gene panel, or chromosome microarray testing within the previous 12 months; speak English or Spanish; and live in the NYC area. Field coordinators at each institution mailed study invitation letters and subsequently followed up via reminder letters and phone calls to parents to determine eligibility, describe the purpose of the study, and schedule data collection. All parents who were present for their child's return-of-results appointment were invited to participate.

We used a stratified purposive sampling approach to ensure a diverse sample of index children stratified by race/ethnicity as recorded in the child's medical record (Black, Latinx, White or multi-racial/ethnic), as well as testing results classification (positive, uncertain, or negative). We successfully recruited a minimum of 5 families per cell, with an extended recruitment period to identify 5 families of children with positive results. Over the course of four rounds of targeted recruitment, we sent letters and made phone calls to 90 eligible families; 22 enrolled, 33 actively declined, and 35 did not respond to recruitment efforts. All 22 families who enrolled also completed the study. The final sample of participants included 24 parents of 22 index children (5 Black; 10 Latinx, 5 White; 2 multi-racial/ethnic) who had received genomic test results.

Data collection

Parents could opt for the interview to be administered in English or Spanish in their home or in a hospital setting. Interviewers obtained informed consent from parents prior to data collection. Interviews lasted approximately 1 hour and were audio recorded. English language interviews were transcribed and then checked for accuracy by the first author, while Spanish language interviews were transcribed, translated into English, and then reviewed by a Spanish/English bilingual interviewer. The two dyadic interviews were conducted using group facilitation techniques, to encourage participation from and interaction between both parents; those transcripts were further reviewed by the original interviewers to ensure participant data were appropriately attributed. Participants received \$40 for their participation in the study as well as direct payment of transportation costs.

The data collection team included doctoral, master's, and undergraduate-level interviewers (one male and three females; including two Spanish/English-speakers) who had no interaction with participants prior to screening, with the exception of scheduling communications. The interviewers received advanced training in interviewing techniques to highlight parents' storied experiences with the genetic testing process (narrative interviewing³⁴) while encouraging breadth and depth within those stories (focused interviewing³⁵). Interviewers followed a semi-structured interview guide with parents to address the following domains: perceived purpose of genomic testing, expectations of results, the return of results process and sequelae, and emotional responses. Following each data collection episode, the interviewer recorded a memo to capture contextual data, personal reflections on the interviewing process, and personal interactions with the participants to inform later analyses.

Analysis

Analysis was conducted by a multidisciplinary team of GCs, a geneticist, and qualitative methods experts trained in public health and sociology. First, an analyst constructed a case-based memo for each parent (or set of parents) to capture individual

families' narratives of their experiences with the testing and return-of-results processes according to *a priori* topic areas and then incorporated interviewer memos to add context. These case memos included quotes that were meaningfully descriptive, impactful, or sparked interest into further areas of inquiry across other cases.

Next, the analysis team participated in weekly meetings to present individual cases, with the goal of identifying repeating ideas emerging across cases in the dataset, which were then discussed, refined, and defined in a preliminary codebook along with *a priori* topics from the interview guide.³⁶ Each analyst independently applied the codebook to the same interview to identify areas of (dis)agreement across coders, refine code definitions, and clarify appropriate identification of emergent themes. Working in pairs (1 GC and 1 non-GC), the analysts then applied the codebook to the same interviews using Dedoose, a qualitative analysis software tool, and discussed discordance and coding rationale until consensus was achieved on current and new code applications per interview. Using grounded theory's constant comparative method, in weekly full-team meetings, emergent themes were discussed both within and across cases until thematic saturation was reached, and the codebook was iteratively updated with higher-order themes.³⁷ Following the coding of all interviews by two analysts, a final round of coding was conducted by the first author (D.W.) to update early code applications with the final coding structure.

Last, a sub-analysis was conducted in which thematic findings that emerged from the whole sample were analyzed separately by racial and ethnic identities of parents. This approach was chosen to explore how diverse groups of families may experience the process of genomic testing and return of results in similar or different ways. Thus, analysts examined each of the major themes associated with parent understanding separately by four categories based on parent-reported race/ethnicity: Black, Latinx, White, and multi-race/ethnicity.

Concurrent to the analyses of participant-generated data, the analysis team engaged in the process of "reflexivity," a common practice in qualitative analysis, in which researchers acknowledge and contextualize their own experiences and perspectives on the topics and data at hand, in order to increase overall credibility of findings.³⁷⁻³⁹ During the analytic process, team members reflexively identified their own interpretation of the data (e.g., through the lens of a GC) in the body of analytic memos, during data checks between analysts, and in full team discussions, to identify individual and collective lenses on emergent findings. Quoted data are presented below with the convention (family ID, parent race/ethnicity, child results classification).

Results

In total, 24 parents (20 mothers and 2 mother-father dyads) were interviewed about the genetic testing process of their children in 22 in-depth interviews. Parents in the sample self-reported as Latinx (42%), Black (21%), White (29%), or more than one race/ethnicity (8%). The two mother-father dyads both identified as White. Offered a choice between English- or Spanish-language interviews, 91% chose English and 9% chose Spanish. Index children's genomic results were negative (45%), uncertain (32%), and positive (23%), as described in [Tables 1 and 2](#).

Elements of understanding

Parents' stories about their child's genomic testing experiences elucidated the different ways that parents absorbed the results, making it clear that the construct of understanding is complex and not unidimensional. Three distinct aspects emerged regarding understanding of genomic test results: (1) comprehension, or knowledge of genetic concepts; (2) significance, which emerges through the process of making meaning of positive, uncertain, or negative results; and (3) implications of the results for child and family health.

Comprehension: knowledge of genetic concepts

Parents said that knowledge of general genetics concepts was important in being able to fully understand their child's test results. Although some reported having basic knowledge about genetics from school or from medical practitioners, gaining this knowledge was described as complicated and out of their reach. Parents were clear that their baseline genetics knowledge was minimal regarding "the chromosomes and the X and Y and those types of things" (F05, Black mother, negative) and that they lacked the foundation to interpret their child's specific test results.

Genetics knowledge was not limited to comprehending general concepts such as chromosomes and genes; it also included how these concepts explained their own child's test result, phenotype, and possible presentation of the condition in the family. Conceptually, inheritance was especially difficult for parents to grasp in the context of this testing.

I don't know how to say it in their language, it's a bit weird; they compare [his father's] stuff with mine, and mine with the boy's, and [his father's] stuff with the boy's as well. (F17, Latinx mother, positive)

Despite the conceptual difficulties, some parents were still motivated to know more about the technical details of their child's test result, hoping to find an origin for their child's illness.

Significance: making meaning of positive, uncertain, or negative results

In the clinical setting, results are commonly classified as positive when there is a demonstrated connection between test findings and child's symptoms of disease, uncertain when it is unclear or unknown if the results can explain all or most symptoms, and negative when there is no known genetic explanation for the condition based on the results. All of the parents correctly labeled their child's test result as compared to the classification in their child's electronic medical record. While parents could accurately assign the correct label to the test result returned, their understanding of the label's meaning was more precarious.

Interviewer: What do you think the negative test result means?

Participant: I don't even know. (F02, Latina mother, negative)

Table 1. Summary statistics of parents (n = 24) and index children (n = 22)

	n (%) or mean (range)
Parent gender	
Male	2 (8)
Female	22 (92)
Parent race/ethnicity	
Black	5 (21)
Latinx	10 (42)
White	7 (29)
>1 race/ethnicity	2 (8)
Child gender	
Male	15 (68)
Female	7 (32)
Child race/ethnicity	
Black	5 (23)
Latinx	10 (45)
White	5 (23)
>1 race/ethnicity	2 (9)
Age of child at testing, years	6.4 (0–15)
Classification of child's test results	
Negative	10
Positive	5
Uncertain	7

The ability to understand their child's test results held value for parents. However, parents differed on how important the results were for their child, and this significance was closely aligned with the classification type (positive versus negative versus uncertain) of the results they received.

Positive results: new understanding can provide relief. Parents who received a positive test result (n = 5) all identified that their child's illness was undeniably explained by genetics, and most provided the exact name of the genetic diagnosis. Most of these parents had traveled through a diagnostic odyssey looking for answers to explain their child's condition. Finally learning the cause for their child's illness was a profound relief, and in the process of making meaning of the results, they furthered their understanding of the disease and possible treatments.

I'm really happy, more so because I know my daughter's disease and I know doctors can take that as a base and even if there's no physical cure, perhaps they said they were going to keep pushing to see if in the future they will find a cure for my daughter, a medication. (F09, Latina mother, positive)

With a named result came the potential for action. While a positive test result still might not change the

course of treatment for the child's condition, it could offer additional avenues for coping with it.

I think that might be a really good idea, is to find a support group for him of other kids. Because it's like you know, he's not the only child. When he heard that he was like, "You know there are other kids that have the same thing like you, and they are living life and growing to be successful." When he heard that, that gave him—that kind of put the sparkle back in his eye. (F16, Black mother, positive)

One mother, however, followed a different trajectory. Her newborn had a positive test result for a rare disease with a poor prognosis. She was able to name her child's disease and also described possible problems that her daughter might face.

I was hoping to learn what more that I could watch out for and also know more, the doctors would know more which medications or interventions were more likely to be successful. (F21, White mother, positive)

Although this mother "felt grateful that her results were definitive," her need to better understand the implications for her daughter's care outweighed the relief she felt about the definitive test result.

Uncertain results: generating confusion. Parents whose child's genomic results were uncertain (n = 7) reported the most difficulty with understanding. Compared to those receiving positive or negative results, these parents expressed a wider variety of ways that the uncertain results created confusion for them. One mother learned that there were multiple irregularities in her son's genome, which did not assemble a coherent picture for her.

I'm not delayed. If I passed that onto him, what does that mean? So, come to find out with the actual duplications of the [named gene] and the extra piece on his X, nothing's missing. Nothing—so they—that's how they explained it to me, it wasn't taken away, it's not missing, it's not broken. It's just some extra on top, it doesn't mean that he's delayed. So, they're not—that's one mystery. I passed something on to my son, I'm not delayed, but why is he? So it's, what is it? (F11, multi-racial/ethnic mother, uncertain)

This mom demonstrated comprehension of her son's test results, accurately recounting the technical information communicated by her son's genetics team, but they did not resonate with her. She had difficulty making meaning from the results and had questions about her child's health. Parents of children with uncertain results were most likely to admit an overarching lack of understanding even when they had comprehended information accurately.

Parents often had questions about their own genetic contribution to their child's illness and actively struggled to understand the degree to which heritability accounted for symptom expression. More generally, some wondered

Table 2. Characteristics of families (n = 22) whose children underwent genomic testing

Family ID	Parent gender ^a	Parent race/ethnicity	Child gender ^a	Child race/ethnicity	Child age at testing (years)	Test type ^b	Results classification
F01	F	Latinx	F	Latinx	11.6	M	negative
F02	F	> 1	M	Latinx	4.0	M	negative
F03	F	Black	M	Black	6.3	M	negative
F04	F	Latinx	M	Latinx	11.8	M	negative
F05	F	Latinx	M	Black	3.1	M	negative
F06	F	White	M	White	14.9	M	uncertain
F07	F/M	White/White	M	White	11.9	M	uncertain
F08	F	Black	M	Black	2.0	M	uncertain
F09 ^c	F	Latinx	F	Latinx	11.3	WES	positive
F10	F	Latinx	F	Latinx	6.0	M	negative
F11	F	Latinx	M	>1	1.1	M	uncertain
F12	F	Black	F	Black	4.4	M	uncertain
F13	F	Latinx	M	Latinx	4.9	M	negative
F14	F	Latinx	M	Latinx	3.9	M	uncertain
F15	F	Latinx	F	Latinx	3.2	WES	positive
F16	F	Black	M	Black	12.9	P	positive
F17 ^c	F	Latinx	M	Latinx	14.2	M	positive
F18	F	Black	M	>1	13.5	P	uncertain
F19	F/M	White/White	F	White	5.2	P	negative
F20	F	>1	M	Latinx	0.3	WES	negative
F21	F	White	F	White	0.1	P	positive
F22	F	White	M	White	1.1	M, P, WES	negative

^aF, female; M, male; F/M, female and male parents both attended the interview.

^bM, microarray; P, panel; WES, whole-exome sequencing.

^cParent interview conducted in Spanish.

if “uncertain” meant that some portion of their child’s symptoms were explained by the genetic finding while other symptoms were not.

I need to know more about whose chromosome, or what’s the symptoms that go with it, because I don’t have these symptoms, but I see him have symptoms....He’s 15 years old, you should know how to do all these things—button your shirt, get dressed in the morning, go in the shower, take your shower, he should be able to do this on his own...I don’t understand. I don’t know if this is part of [his genetic condition]. It might be, it might not. I don’t know. (F06, White mother, uncertain)

Many parents entered the testing process with expectations that the test would yield answers about their child’s health and prognosis, despite pre-test counseling efforts to prospectively manage them. When expectations were unmet, parents were overcome by their dashed hopes (a description of these dynamics are described elsewhere⁴⁰). Uncertain results therefore left parents both disappointed and unable to make meaning of the results.

I thought I was going to see bad stuff, if he was going to have in the future, disabilities. But so far so good, they gave me good news. But they said they want to do more testing, so that got me confused. (F14, Latinx mother, uncertain)

Negative results: satisficing with minimal understanding.. Parents who received negative genomic test results (n = 10) exhibited a behavior commonly understood in the social sciences as “satisficing.” Satisficing occurs when people reduce their actions from optimal to acceptable, or merely satisfactory. The investment to maximize the situation is not warranted, because the stakes are not high enough.^{41,42} In the context of genomic testing, if the child received a negative test result, most parents satisficed, accepting an incomplete understanding of their child’s results because they did not feel they needed to pursue it further—basically, the test result had not changed anything. Parents described that receiving a negative result was good enough, they saw little benefit in trying to make further meaning of results, and therefore they were comfortable ending their pursuit for a genetic explanation for their child’s condition.

Yeah, they sent me all the paperwork with the results. I mean, I couldn't read it. I just knew that every single page just had like, "Negative" on it and that's all I really cared about....If there was a positive finding I think it would've been helpful, but because it was negative I didn't want to read through what it was because it didn't matter. (F22, White mother, negative)

However, when probed further by the interviewer about the meaning they assigned to the negative result, confusion was a common experience.

It's like, no, nothing's wrong, but then it's like you start thinking about it. It's like, oh, so what now? (F05, Black mother, negative)

Some even conflated a negative test result with the absence of disease. They described the results as "normal" or "good" and even wondered if their child was being declared healthy by testing negative.

Yes, [the counselors] were clear with the results. I understood that she's normal, that there's no concerns....I mean they explained that everything that they tested for was normal. (F01, Latinx mother, negative)

A minority clearly described how they were asked to stay in touch with their genetics providers in case more was learned in the future that held potential for explaining their child's condition.

Well, I repeated it like three times and everything comes back fine, so it's like I think last year I didn't do it. I think they want to do it this year. They say it's good to do it because every year there's different testings. (F13, Latinx mother, negative)

This was a particularly confusing message to parents who had received negative test results, because most interpreted a negative result as being "cleared" from any genetic explanation for their child's condition. Being asked to return in the future to look for a new genetic result conflicted with their satisficing heuristic.

Implications for child and family health

By far the most problematic part of genomic testing for parents—almost regardless of the test results themselves—was how to contextualize the implications of the results for the health of their child and their family. For many, a satisfactory understanding was achieved only when they could apply the test findings specifically to their child. One mother, whose child had received a negative result, longed for a solution to her daughter's pain symptoms.

We don't know exactly why it's happening, but we know it's this at least, and we can help her with pain management and that's what's kind of more important to me, because she was in a lot of pain for quite—like six months, so I just—that's what's kind of my main priority. Anything to kind of help her with that pain. (F10, Latinx mother, negative)

Parents wanted to understand more about their child's diagnosis and prognosis and whether there were future treatment options. That meant sometimes imposing meaning on the test results so they could gain clarity about straightforward clinical utility.

I think that everybody involved wants some definitive answer that we can point to and say, "That's what's been happening and here's how we fix it," and the problem will go away." (F19, White father, negative)

Understanding testing implications was not limited to the index child but also included how the results might affect other family members, such as whether genetic factors could have implications for siblings or future generations.

We have another son, so we just want to be sure for our son when he's growing up and might want to have kids. If it's a genetic thing, we just want to let him know about this thing (F07, White father, uncertain)

Parents felt that understanding their child's current disease diagnosis and management could make it easier to know how to handle a future case in the family. In these circumstances, parents were reflective on both clinical and personal utility for future generations.

[The doctor] said this to [my son]. "Better that you know that you have it, so now before the doctors didn't know to look for it. Now when YOU have a kid, now the doctors can go testing, to see if your son or daughter has it." If it ever comes to the point where [his] child has that, at least they have somebody to relate to. (M16, Black mother, positive)

Processing information, assembling understanding

Genomic test results involve a great deal of information, which can be complicated for parents to process. Parents engaged in two complementary approaches to actively assemble meaning from the results: outsourcing some of the work to providers and seeking out internet resources. By "assemble," we mean that parents took the different parts of the information and the different ways of understanding and joined them to form a coherent story.

Outsourcing genetics knowledge: providers are custodians of the details

Regardless of results classification, parents overwhelmingly accepted that they had a minimal level of technical knowledge about genetics concepts generally and their child's test results specifically. Parents explained how their providers attempted to share complex information with them, yet how little of it they absorbed or perceived that they understood.

They always have their technical words, but I expected it, they're doctors. [laugh]. They have their big words. But if they—they do it in a form that they break it down, I can understand. (F10, Latinx mother, negative)

Parents generally were not bothered by their lack of comprehending details. Instead, they expected their providers,

who were viewed as having much more expertise than parents to comprehend the technical results, to be *de facto* custodians of the information, and, therefore, they sometimes refrained from asking clarifying questions.

We were lost a little bit...[the doctor] was really pleasant. We just smiled and went through it. It's just a little awkward. It felt like we were listening to someone speak in another language (F04, Latina mother, negative)

But they know how to do their things. They're doctors. You know, I just get guided by what they tell me what to do with him. (F02, Latinx mother, negative)

Parents appreciated the work that providers did for them to explain the detailed technical results but were quick to move beyond wanting to understand providers' technical explanations. While providers dwelled on communicating technical information about genetics and specific test findings, parents were more in favor of understanding implications for their child's health.

And then they told me they found this, they found that, but then I'm like, "Is that something that should concern me?" (F01, Latinx mother, negative)

In fact, some parents felt overloaded by the complexity and amount of technical information that providers tried to impart during the return of results and preferred less of it.

When you go to get the results they actually tell, "Do you remember you came for this and this and this?" and they tell you a whole bunch of different numbers and stuff like that. They go into it detail by detail like sometimes you're like, "Okay, I already went through this. Just give me the results." And they're like, "No, we got to go through it." (F13, Latinx mother, negative)

Although there was sometimes a mismatch between parents and providers about the importance of fully comprehending results, parents predominantly described their experiences with providers benevolently. Parents highlighted many ways in which their providers sincerely cared for the well-being of their child and family.

I feel like they've been really human with me, which is to say that they're not just looking at this like—they're not treating—they're not just focusing on the illness. They're focusing on our family, or on our personalities, on our process. (F21, White mother, positive)

Some highlighted the amount of time, unrushed, that providers spent with them during the testing process and return of results. Regardless of the outcome, parents lauded their providers' efforts to optimize their understanding and viewed provider efforts as an additional expression of care that held meaning for parents.

They tell you what's going on. They don't leave nothing out. They ask you, "Do you understand?" Like, they

keep it open to want to know if you're okay, if there's anything you would like to know. They keep it going for you to be better understanding about what's going on. They care about you. They care about your child. Because they know this is a delicate situation. (F03, Black mother, negative)

"Don't read that": contending with online resources

Outsourcing knowledge of the technical results to providers was a process that parents often did in the clinical space, but sometimes parents turned to online resources to supplement what they did not comprehend or absorb from their clinical experiences. Sometimes these resources proved helpful in providing context or reinforcing provider messages.

I did go and search on the internet and what [my providers] explained to me, that's what I found—the same thing. (F09, Latinx mother, positive)

More often, however, parents reported emotional costs associated with turning to internet sources to improve their understanding of their child's condition or test results. As parents attempted to assimilate information about their child's condition following clinical visits, a variety of emotions surfaced, including sadness, anxiety, and fear. These emotional responses sometimes fueled parents to seek out information from internet sources in an effort to reduce these difficult feelings. One mom reflected on her own information-seeking behavior.

If you don't understand something you start being scared. They tell you all the time do not look at WebMD, don't read that. Because you're going to read something and you're not going to understand it and you're going to automatically think I have that. (F11, multi-racial/ethnic mother, uncertain)

Ironically, as parents tried to satisfy their desire for understanding vis-à-vis internet resources or social media, these outlets typically exacerbated difficult emotions. In some cases, parents acknowledged that when they began to feel strong emotions of overwhelm and fear, they discontinued information seeking entirely. Specifically, parents became aware of their percolating emotions and self-protected by avoiding internet searches unmoderated by their care providers.

I went on YouTube trying to search it and there was one that wasn't a good story like it was a heartbreaking story. So then I just stopped. I was like those are sad stories that you know not everybody is the same so I can't just focus on something like that happening. I was like everybody's story is different. (F02, Latinx mother, negative)

Overall, we did not identify any differences in the ways that Black, Latinx, White, or multi-racial/ethnic parents described their understanding of their child's genomic test results in any of the above themes.

Discussion

This research provides important insights into the meaning and process of understanding genomic test results among a sample of racially and ethnically diverse parents. In describing their experiences with their child's genomic testing process, parents identified multiple dimensions of understanding their child's test results: comprehension or having knowledge of technical genetic concepts, significance and making meaning of results according to their classification, and identifying implications for their child's health and family's future.

Understanding clinical utility and family implications is a priority

Parents in this study were quick to label genetics knowledge as a relevant component for understanding the particulars of their child's results. However, they did not feel that developing this type of knowledge was within their own reach. They easily assigned this custodial role to providers who could harbor the technical knowledge and details. This precluded parents' need to develop their own genetics knowledge—not a primary goal for parents. This is a critical observation.

Many studies on the effectiveness of genetic counseling strategies measure genetic and genomic knowledge as a desirable outcome in lieu of understanding or perceived understanding.^{12,43,44} From the parents' perspective, however, developing this knowledge was not a priority relative to the implications for their child's and family's health. Overwhelmingly, parents wanted to understand the health implications of the results for their child—the clinical utility of the results. This is consistent with previous findings that patients preferred their providers to expressly state the clinical utility of the genetic test results and that counseling addresses personal meaning for the patient and their family.^{21,45} Understanding potential implications for the rest of the family was a related priority, aligning with others' descriptions of elements of personal utility, such as consideration of reproductive autonomy or the ability to plan for the future outside of clinical concerns.⁴⁶

Problems with labeling results as positive, negative, or uncertain

Current categories of genomic test results may be useful labels for clinicians, but they may confuse parents more than they help. In some instances, a negative result inappropriately lulled parents into dismissing the possibility of a genetic explanation and claiming all was normal, despite the possibility that emerging science over time might find a genetic cause. In one study examining the return of negative results, most patients were confident they understood their results, but only 20% actually had a nuanced understanding of what negative results actually mean.⁴⁷ The negative test label may unintentionally reinforce parents' misconception that the results definitively

ruled out a genetic explanation for their child's condition or even that their child did not have a medical condition at all.

Additionally, it has been shown in previous studies that uncertain results can increase confusion and misunderstanding among patients receiving genetic testing for a variety of indications and among parents of pediatric patients.^{48–51} Clinicians may want to consider renaming these classifications to match the way parents receive the information. Suggested terms include “not positive”⁴⁷ or “uninformative” to encompass both negative and uncertain result categories together, but these will need further testing to ensure their appropriateness.

Delivering test results: reflections on roles and responsibilities of GCs

This analysis benefitted from in-depth reflexivity activities to identify how context and the analysts' own profiles shaped interpretation of the data and, ultimately, the study findings. When GCs on our research team (authors J.A.O., S.A.S., K.M.G., and K.E.D.) analyzed interview data about parents' understanding of their child's test results, they identified a disconnect between their own standard of practice—comprehensive understanding—and the goals of parents, which were more limited. Specifically, GCs reported feeling a responsibility to deliver robust genetic information as part of their practice, with the goal of maximizing parent understanding of the results. In contrast, we found that some parents' primary need for understanding was not comprehension—it was much more narrowly focused on implications for their child and family. This finding highlighted the discomfort that GCs have about “rationing” or limiting information clinically. They hold professional values that dictate that they are ethically and legally required to deliver comprehensive information to the parents who are responsible for making health care decisions.

Although contracting is an important component to genetic counseling practice, there may be limitations on the degree to which GCs can alter the amount and type of information about the genetic test results they disclose to the patient or parent. This poses the following conundrum: GCs believe they are responsible for disclosing specific, prescribed details about the results, and although they can provide additional details if the patient desires, they are uncomfortable withholding what they see as essential education. For example, if a result has potential implications for other family members, is it necessary for GCs to explain complicated concepts about inheritance, even if the parent does not grasp or does not find value in hearing the information?

The GCs who participated in this analysis reflected on the standard approach to returning genomic test results to parents as being information-heavy by default and questioned the effectiveness of this approach toward the goal of parent understanding. Potentially, this information-heavy approach to genetic counseling is explained by vestiges of “genetics exceptionalism,” a concept that singles out

genetic findings as distinct from other clinical findings and therefore deserving a different, and highly complex, approach to returning results.^{52,53} In other fields of medicine, patients have reported preferences for a provider-driven approach^{54,55} and more recently even in genetics research.²² GCs considered whether a more direct approach to counseling for some parents (i.e., one that prescribes a stronger focus on clinical and personal utility and less on genetics knowledge) might better meet parent expectations, improve self-perceived understanding, and perhaps even increase overall satisfaction with genomic testing.⁴⁰ Our research raised these important questions as part of our analyses of parent understanding of test results, but implications for genetic counseling may stretch beyond this topic and deserve further exploration. Findings from ongoing research projects within the CSER consortium that are investigating alternative result disclosure models, including a literacy-focused method³² and a digital platform that allows parents to control the amount and flow of information,⁵⁶ may help inform methods for adopting a more personalized approach.

As patients have increased information-seeking behavior on the internet, healthcare providers have assumed new roles as interpreters of this online information.⁵⁷ Additionally, physicians have recognized that the use of online resources can increase distress and misunderstanding of medical information among patients.⁵⁷ GCs regularly assume the role of internet interpreter, which diverges from their traditional role as information provider.¹ We found that some parents turned to online resources to help process information and therefore assemble a coherent understanding of their child's genomic test results. Yet for some, this information-seeking was counterproductive, intensifying negative emotions. These findings suggest that GCs have an opportunity to address parents' psychological and social needs in post-return-of-results communications both to minimize negative feelings from online engagement and to address further questions. In this way, GCs are uniquely positioned to respond to parents' internet-generated questions, ensuring parents clearly understand any related implications for their child or family.

A call to explore genomic testing experiences among parents of diverse racial and ethnic backgrounds

Finally, when we conducted a sub-analysis of themes stratified by race and ethnicity, Black, White, Latinx, and multi-racial/ethnic parents all understood their children's test results in similar ways to one another. This finding adds important evidence to the minimal body of research documenting ways racially and ethnically underrepresented parents understand their child's genetic test results. In this case, elements of understanding were similar and similarly prioritized, regardless of race or ethnicity. Other research has found that speaking English as a second language can be a barrier to parent understanding;²² however, our limited data from the two parents who spoke English

as a second language did not suggest this. While our findings about parent understanding in this study did not differ by general categories of race and ethnicity, we recommend additional inquiry into these topics along with questions about the impact of level of education and language on understanding. Further, as access to genomic medicine expands, it is increasingly important to explicitly explore the ways parents from diverse racial and ethnic backgrounds experience clinical encounters differently, to address the potential for implicit bias among typically White GCs, as seen in other genetic research.^{58,59} Recognizing the homogeneity of White women in the genetic counseling profession and the importance of diversity, equity, and inclusion (DEI), the National Society of Genetic Counselors (NSGC) released a DEI position statement acknowledging historical injustices, advocating for underrepresented people within and served by the profession, and valuing difference as a guiding principle. NSGC has launched multiple DEI initiatives to support its position, including a DEI Advisory Group, a Cultural Representation and Outreach subcommittee, and coursework designed to help counselors recognize and address implicit biases.⁶⁰

Limitations

We present findings from parents whose children received test results from various providers in two clinical care settings. Parents did not always distinguish experiences between different types of providers: geneticists, GCs, or other medical professionals. This made it difficult to distinguish methods of practice between provider type, if any existed. The benefit of exploring these experiences in a clinical care setting (versus a standardized research setting) meant that these experiences may have more real-life applicability. Additionally, index children underwent different types of genomic testing. It is therefore possible that different types of testing may be more or less difficult to explain and for parents to understand. These potential differences were not explored.

Some parents were interviewed within weeks of receiving results, while others had received them nearly one year prior. It is possible that recall errors occurred, although all parents accurately reported their child's genetic test results, and responses to questions about understanding did not appear to vary systematically by length of time since the test. All interviews except two were conducted in the hospital where their genetics care had been provided. While interviewers explained during the consent process that interview data were confidential and would not be shared with providers, it is possible that participants may have self-censored negative experiences for concern over interviewer affiliation. Further, the two families that participated in dyadic interviews may have co-produced data that were different than those produced by individuals interviewed (i.e., more succinctly detailed due to interactional clarification). However, due to the small number of dyadic interviews conducted, it is not possible to evaluate what the different modes of data collection may

have produced as attributable to the method versus unique family experiences.

Conclusions

In this study, comprehension—knowledge of technical genetic concepts—was not important to parents. These were little more than background or context, facts without meaning. Instead, parents primarily wanted to understand how test results might affect their child's care and family's future, relying on their providers to manage the complicated details. Despite the perceived best efforts of providers, most parents lacked confidence in their understanding of test results, leading them to reach for online sources that were less than helpful. Parents also achieved different levels of understanding depending on the type of results they received, with some (positive, uncertain) placing more value on it than others (negative). Additionally, the classification terms assigned to genomic test results, in and of themselves, misguided parents. Finally, reflexively analyzing these data through the lens of genetics counselors identified a mismatch between a provider-driven, information-heavy practice in disclosing genomic results and parents' preference for acquiring clinically relevant meaning for their child and family.

GCs work diligently to communicate what they think parents need to know in order to understand their child's results, and parents generally appreciate the effort. However, as genomics moves into the mainstream, and counseling efforts will inevitably need to be streamlined, perhaps it is time to emphasize what parents really seek to understand: how to best care for their child and family.

Data and code availability

There are restrictions to the availability of interview data due to patient privacy or ethical restrictions.

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Declaration of interests

The authors declare no competing interests, with the exception of Eimear E. Kenny, who received a speaker honorarium from Regeneron Pharmaceuticals and Illumina.

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