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ORIGINAL ARTICLE

Parents' preferences for receiving and discussing prognostic genetic information regarding their children's neurodevelopmental condition: A qualitative study

Erin Turbitt[1] | **Meg Bourne**[1] | **Alison McEwen**[1] | **David J. Amor**[2,3]

[1]Discipline of Genetic Counselling, University of Technology Sydney, Ultimo, NSW, Australia

[2]Murdoch Children's Research Institute, Parkville, VIC, Australia

[3]Department of Paediatrics, University of Melbourne, Parkville, VIC, Australia

Correspondence
Erin Turbitt, Discipline of Genetic Counselling, Graduate School of Health, University of Technology Sydney, Building 20, Level 6, 100 Broadway, Ultimo, NSW 2007, Australia.
Email: erin.turbitt@uts.edu.au

Abstract

Aim: To investigate parents' preferences and motivations for receiving and discussing prognostic genetic test results.

Method: We used a cross-sectional, interpretive description qualitative study design. We collected data through semi-structured interviews with Australian parents, which we analysed using reflexive thematic analysis.

Results: Parents ($n = 32$) had a child or children with a genetic neurodevelopmental condition, such as fragile X syndrome, DiGeorge (22q11.2 deletion) syndrome, or Angelman syndrome. Parents of mildly impacted or older children were tolerant to prognostic uncertainty. Parents found conversations about their child's prognosis emotional and preferred to discuss their child's potential strengths and challenges. While most were enthusiastic about prognostic tests and described many motivations for testing, the potential for prognostic information to contribute to a loss of hope and stigmatizing societal views were also discussed.

Interpretation: Parents had mixed preferences and motivations for acquiring prognostic genetic information about their child, contrasting evidence in other contexts such as cancer where parents typically have minimal tolerance of uncertainty. Health professionals should consider strength-based framing of prognostic information gained from current and emerging technologies when returning results to families.

Neurodevelopmental conditions are the most frequently encountered chronic conditions in paediatrics.[1] Genetic investigations are recommended to identify an underlying diagnosis for children presenting with intellectual disability, global developmental delay, or autism.[1] Efforts to explore the addition of genetic tests to newborn infant screening programmes are under way.[2] Consequently, paediatricians are increasingly tasked with disclosing genetic diagnoses to families.

Genetic neurodevelopmental conditions encompass a broad spectrum of conditions arising from spontaneous (de novo) or inherited gene variants. They include conditions such as fragile X, Angelman, and DiGeorge (22q11.2 deletion) syndromes.[3] While there are distinct differences between the phenotypes of each condition, all are characterized by cognitive and behavioural impacts and have a paediatric onset.

Conversations where genetic test results are disclosed generally include discussion of the expected prognosis for the child.[4, 5] At present, most genetic tests are diagnostic, providing a yes or no answer as to whether a child has the condition. Genotype–phenotype correlations may provide some indication of expected prognosis based on diagnostic genetic test results that is personalized to an individual; however, most conditions vary greatly in terms of prognosis within their diagnostic category.[6] Thus, discussions about the expected prognosis from diagnostic genetic tests are not tailored to the specific child but based on scientific data (or clinical experience) about the expected range within the diagnostic category.

Meanwhile, new 'prognostic' tests (such as those based on the epigenetic profile of an individual) are being developed for some neurodevelopmental conditions. These prognostic tests promise more precise prognostic information, including the ability to predict the degree of disability expected for the child. For example, in fragile X syndrome, a test has been developed based on evidence that increased methylation levels (a type of epigenetic signature) are associated with lower intellectual functioning and greater behavioural challenges in childhood.[7,8] It has been proposed that such a test could be used to provide families with more precise prognostic information.[8] The test can be performed on blood spot samples, offering the potential for use in newborn infant screening, well before symptom onset.

With these new tests on the horizon, it is critical to ascertain the views of families regarding prognostic information. Much of our understanding about patient preferences for prognostication comes from oncology, where prognostic information is important for patient decision-making about treatment, or to prepare for end of life.[9] Patients with cancer (or their parents) report a preference for honest and transparent prognostic disclosure.[10,11] Similarly, parents of children in the neonatal intensive care unit value concrete information about their child's anticipated abilities and potential limitations.[12]

The generation of this evidence regarding patient preferences has led to a paradigm shift whereby clinicians traditionally censor information indicating poor prognosis in an attempt to retain hope for families;[13] yet, as described earlier, patients value honesty,[9] which allows patients and families to reassess what they hope for, rather than removing hope altogether.[14] Clinical guidelines similarly recommend that clinicians have honest conversations with families about prognosis as it pertains to end of life.[15]

Uncertainty is common in paediatrics, particularly where clinical genetics and prognostication intersect. Uncertainty in illness theories provide a useful backdrop for understanding parents' preferences and views about prognostic genetic tests.[16] Uncertainty is a meta-cognition experienced when an individual is consciously aware of their ignorance, and historically has been treated as a pathological disorder to be eradicated through actions such as genetic testing. However, there is growing support for uncertainty to be conceptualized as a normal state of being to be accepted and managed.[17] A person's ability to manage such uncertainty is referred to as 'tolerance of uncertainty'.[18]

The taxonomy of medical uncertainty proposed by Han et al.[19] delineates different sources of uncertainty that are useful to consider in the context of genetic testing, including probability and ambiguity. Probability refers to uncertainty about the risk of a given outcome, whereas ambiguity refers to uncertainty regarding the validity of risk information. In the case of prognostic information, ambiguity could refer to imprecision of test results because of conflicting scientific evidence.[19]

Despite this robust evidence base and theoretical understanding, we suggest that there may be an important nuance to how parents of children with neurodevelopmental conditions prefer to learn about and discuss prognostic information related to cognition and behaviour that has not been previously explored.[9] Most previous research on prognostication focused on conditions that are life-limiting, and predominantly considered physical health outcomes with less attention given to neurological outcomes.[9] Our focus on prognostication regarding cognitive and behavioural aspects in conditions that are chronic and not life-limiting is new and timely given the evolving societal context of promoting acceptance of neurodiversity.

What this paper adds

- Parents had varied views about receiving prognostic information on their children's neurodevelopmental condition.
- Some parents preferred prognostic uncertainty about their children's genetic neurodevelopmental condition.

We sought to extend the exploration of chronic illness prognostication by understanding parents' experiences with learning and discussing prognostic information about their child's genetic neurodevelopmental condition. We further aimed to explore parents' preferences and motivations for receiving more precise information from a prognostic genetic test.

METHOD

Study design

We used a cross-sectional, interpretive description qualitative study design,[20] collecting data through semi-structured interviews and analysing data using reflexive thematic analysis (see the data analysis section for further details).[21] We adopted a contextualist stance because we wanted to study people in the context of their lives and acknowledge multiple accounts of reality.[22] In using contextualism, we allowed our values and practices as researchers to shape the collection and analysis of data. This meant that our experiences and perspectives were combined with those of participants to co-create meaning, rather than attempting to account for or remove our influence as researchers. For example, we made judgements about the usefulness of data as it related to answering our research questions and contributing new knowledge, through our clinical expertise and knowledge of current evidence.

We reflected on our subjectivity and the impact on the collection and interpretation of data. ET is a social scientist and experienced qualitative researcher whose work focuses on parental decision-making, with experience working in molecular genetics laboratories. AM is a genetic counsellor and educator. DA is a clinical geneticist. Both AM and DA have experience providing care for families of children with

genetic neurodevelopmental conditions. MB was a Master of Genetic Counselling student at the time of conducting the study. We do not have lived experience parenting a child with a genetic neurodevelopmental condition.

Our research was informed by the literature and theory, which highlight the role of uncertainty in health judgement and decision-making. We used theory to shape our research questions and interview guide, and to direct and facilitate data analysis and theme development.

Participants and recruitment

We used purposive sampling to recruit the parents of a child with a diagnosed genetic neurodevelopmental condition. We posted study advertisements on the Facebook pages and in newsletters for Australian support organizations, including the Fragile X Association of Australia, Angelman Syndrome Association Australia, and Rare Voices Australia. Interested parents contacted ET via a link to a REDCap survey or direct e-mail.

We used the information power model to determine the number of participants.[23] This involved consideration of five parameters (study aims, sample specificity, use of theory, quality of dialogue, and analysis strategy) to determine how many participants were needed to ensure that the sample holds sufficient information to answer the research question. Our narrow study aims and use of theory indicated that fewer participants were required. However, we included the parents of children with a range of conditions varying in age, which increased the required number of participants to reach sufficient information power. Information power was evaluated throughout the study. Because of the varied quality of dialogue in some instances, we again required an increased number of participants. These factors combined led us to determine that a sample of a maximum of 32 participants would be sufficient to reach information power and answer the research questions. On determining this sample size, we secured funds to offer a A$50 gift voucher to 30 participants to acknowledge their time (two participants opted not to receive a voucher).

Data collection

ET conducted the interviews between July and September 2021. We gave parents a choice to be interviewed over Zoom (with or without video), telephone, or e-mail. We developed the interview questions using input from advocacy group leaders, clinicians, and researchers. We piloted our interview schedule with two families of children with fragile X syndrome, who provided feedback on the questions and the overall focus of the interview.

Our interview schedule (Appendix S1) covered questions around gaining information about the child's condition and diagnosis, experiences with genetic testing, including receiving diagnostic and prognostic information (e.g. Can you tell me about any information you've received about how your child is likely to develop as they get older?), their thoughts on prognostic genetic testing (e.g. Would you be interested in information that tells you whether your child may be more or less severely affected compared to the average child with that condition [either at the time of diagnosis or further along]? Why/why not?), and decision-making processes (e.g. How would you go about making the decision to get your child tested?).

We collected the key characteristics of the parents and their children during the interviews. This included parents' location (their state and metropolitan or regional area), level of education, and ethnicity. We asked parents about their child's genetic diagnosis, current age, and age at diagnosis. To understand the impact of the condition on their child, we categorized the information parents provided as mild, moderate, or severe. This categorization was based on parents' descriptions of their child's condition (e.g. 'he is severely disabled') or how they compared their child's functioning to other children with the same condition (e.g. 'she is probably on the higher level of what people with Angelman syndrome can do').

Data analysis

We audio-recorded the interviews, transcribed them verbatim using the Rev automated transcription services, and checked them for accuracy. We used reflexive thematic analysis.[21] ET coded the data, highlighting any sections of the transcript that would contribute to answering the research questions. This involved both semantic and latent coding. We used semantic coding to identify explicit or surface-level meanings and latent coding to go beyond the descriptive level, interpreting meaning based on our knowledge of the topic and theory. While ET was the sole formal data coder, all members of the research team had access to the transcripts and we met regularly to discuss the preliminary ideas and concepts being developed. The purpose of these meetings was to deepen our understanding of the data and explore multiple interpretations, rather than to ensure 'consistency' or 'reliability' of coding as these concepts align more with a positivist paradigm.[24]

Themes were developed from codes by compiling and organizing them to identify broad patterns in the data that were relevant to the research questions. ET used a visual mapping technique to determine how themes related to each other and develop the overall story. These candidate themes were reviewed and revised with DA and AM to determine the final themes and ensure that the analysis was useful and relevant to the research questions.

Ethical statement and consent

The project was approved by the Royal Children's Hospital Human Research Ethics Committee (HREC) (no. 69604) and ratified by the University of Technology Sydney HREC (no. ETH21-5714). All participants provided verbal consent as per the HRRC-approved protocol.

TABLE 1 Participant characteristics.

Characteristic	*n*	%
Child's condition		
Fragile X syndrome	9	28.1
DiGeorge syndrome (22q11.2 deletion)	5	15.6
Angelman syndrome	5	15.6
Other rare deletion and duplication syndromes	5	15.6
Phelan–McDermid syndrome	3	9.4
15q11-13 duplication	2	6.3
Mosaic Down syndrome	1	3.1
16p11.2 deletion	1	3.1
Sotos syndrome	1	3.1
Child's age at the time of the qualitative data collection		
<2 years	2	5.7
2–5 years	13	37.1
6–10 years	8	22.9
11+ years	11	31.4
Unknown	1	2.9
Age at diagnosis		
Prenatal	2	5.7
<1 month	4	11.4
1 month to 2 years	19	54.3
>2 years to 3 years	5	14.3
>3 years	5	14.3
Time since the diagnosis		
<6 months	3	8.6
6 months to 2 years	8	22.9
>2 years to 3 years	9	25.7
>3 years	15	42.9
Location		
Metropolitan	22	68.8
Regional	5	15.6
Unknown	5	15.6
Parent described the impact of the condition on the child as …		
Mild	7	21.9
Moderate	11	34.4
Severe	11	34.4
Child too young to determine	3	9.4
Location (state)		
New South Wales	12	37.5
Victoria	10	31.3
Queensland	4	12.5
Western Australia	2	6.3
Tasmania	1	3.1
Unknown	1	3.1
Based overseas	2	6.2
Parents' highest level of education		
University or tertiary institution	19	59.4
Secondary school (range: 7–12 years)	6	18.8
Technical or further education institution	5	15.6
Unknown	2	6.2
Parents' self-reported ethnicity		
Australian/New Zealand European	24	75
Indian	2	6.3
Coptic Egyptian	1	3.1
European	1	3.1
Italian/South American	1	3.1
Maltese	1	3.1
Unknown	2	6.3

RESULTS

Participant characteristics

Fifty-five parents expressed interest in participating in an interview. Five of these parents made contact after the recruitment target was reached and were offered the option of being contacted in the future to participate in similar research. Of the remaining 50, 32 participated in the study, four were unable to schedule a time for an interview, and 14 did not respond to e-mails inviting them to participate in the study. No parents were excluded based on the eligibility criteria. The average length of the interviews was 34 minutes (range: 17–56 minutes). Of the 32 parents, 16 participated via telephone, 12 via Zoom (with video), and four via e-mail.

Participant characteristics are shown in Table 1. Most participants (59.4%) had completed education from a university or tertiary institution, 18.8% from a high school, and 15.6% from a technical or further education institution. Most participants ($n = 24$, 75.0%) identified as Australian or New Zealand European and were female ($n = 31$, 96.9%).

Nine (28.1%) participants had children with fragile X syndrome, five (15.6%) with DiGeorge (22q11.2 deletion) syndrome, and five (15.6%) with Angelman syndrome. Other conditions are listed in Table 1, some of which are not named to maintain the confidentiality of families. Three families had more than one child with fragile X syndrome. Over half (60%) of the children were diagnosed before 2 years of age or prenatally; all were diagnosed before turning 10 years of age.

Themes

We developed four themes, all of which are potential determinants of preferences for prognostic information (summarized in Table 2).

TABLE 2 Theme summaries.

Theme	Central organizing concept and scope
1. Acceptance of uncertainty (some parents seek prognostic information to reduce uncertainty, while others have a preference for prognostic uncertainty).	Tolerance to uncertainty about their child's future can change over time with parents generally becoming more tolerant as their child ages. Parents are comfortable with potential inaccuracies about prognoses (ambiguity). Uncertainty and ambiguity may be preferable, providing hope for the child's future potential. *Subtheme: Parents' acceptance of their child's condition helps acknowledge the irreducibility of uncertainty*
2. Psychological needs and coping responses (conversations and decisions about prognostic information are emotional for parents, indicating the importance of psychological support).	Parents experience negative emotional reactions to learning prognostic information. Provision of balanced, strength-based framing of prognoses was recommended.
3. Decision-making and motivations to receive prognostic information (parents are enthusiastic if the information will help with practical and emotional coping, although they stressed the importance of informed choice about prognostic testing).	Parents are motivated to receive prognostic information to help with practical and emotional coping. *Subtheme: The decision-making process about undergoing prognostic genetic testing (a risk–benefit analysis)* The potential outcomes (benefits and risks) of receiving prognostic results inform parents' decisions about the use of medical interventions, including genetic tests.
4. Parents' acknowledgement of possible societal harms of more precise prognostic information.	Reflections about the potential unintended consequences for society of prognostic information being available.

Theme 1: Acceptance of uncertainty (some parents seek prognostic information to reduce uncertainty, while others have a preference for prognostic uncertainty)

Theme 1 captures the role of uncertainty (explored as both probability and ambiguity) on parents' interaction with prognostic information. Parents described wanting or searching for prognostic information to reduce uncertainty early in the diagnostic process (Table 3, quote 1).

Overall, parents of children who were more mildly impacted appeared more tolerant of prognostic uncertainty compared to parents of more severely impacted children (Table 3, quote 2). Parents discussed how their tolerance to uncertainty changed over time, from initially being intolerant to uncertainty, to more tolerant as their child aged. Some described developing a preference for an uncertain, more open future for their child.

Many parents described a need for certainty at the time of the diagnosis and expressed feeling disappointed at the lack of prognostic information given by health professionals who provided the diagnosis or were involved in their child's care (Table 3, quote 3). Those searching for prognostic information discussed learning from families of older children with the same diagnosis. These parents compared the type of genetic variation using this information to form their own predictions about their child's future (Table 3, quote 4).

There was generally a high level of comfort with ambiguity among parents as it related to the potential inaccuracies in prognostic information provided by new genetic tests. Parents acknowledged that no medical test is 100% accurate (Table 3, quote 5). Those who acknowledged prognostic ambiguity often brought up environmental influences and early intervention, which would not be accounted for in generating predictive prognostic information (Table 3, quote 6).

Parents discussed that ambiguity about test results could be beneficial. In the case of an unfavourable result, indicating a more severe prognosis for their child, parents thought that the possibility of inaccurate test results would give them a chance to 'prove them [health professionals] wrong' and the results would be less likely to limit their child, compared to being provided with a result that is guaranteed to be accurate.

Subtheme: Parents' acceptance of their child's condition helps acknowledge the irreducibility of uncertainty

Parents' interest in receiving more precise prognostic information was tied to their level of acceptance of their child's condition. Those who could readily describe their child's strengths and positive aspects of their child's condition were generally less enthusiastic about the prospect of learning more specific prognostic information. Parents explained that with or without this additional information, their child would still be the same person and they would support their child as best they could regardless (Table 3, quote 7).

The availability of more precise prognostic information was predicted to help parents accept and come to terms with their child's condition (Table 3, quote 8). Conversely, others discussed that it would be more appropriate to provide more precise prognostic information after the diagnosis once parents had a chance to adjust and accept their child's condition (Table 3, quote 9).

Theme 2: Psychological needs and coping responses (conversations and decisions about prognostic information are emotional for parents, indicating the importance of psychological support)

Parents described the time of diagnosis and receiving associated prognostic information as overwhelming, stressful, and

TABLE 3 Illustrative quotes: Theme 1: Acceptance of uncertainty (some parents seek prognostic information to reduce uncertainty, while others have a preference for prognostic uncertainty).

In-text quote number	Verbatim quote
1	'We do want to know whether she's on there more affected side or the less affected side. That is something that we want to know, and that is something we've actually asked for and there's no way of knowing that yet.' Parent 004 (female child with 22q11.2 deletion, 7 months old, 6 months since diagnosis)
2	'And [the paediatrician's] advice at the time was probably quite good because they said, 'I don't know where he is going to be', and they met [my son], so they did some initial assessment. [My son] was in the room with us, [the paediatrician] said, 'look, where he's at now he's in a good place. We'll just have to see how it goes. If you've got any other questions, come down again'. So, they weren't very specific, but it was actually a lot more helpful.' Parent 007 (male child with 15q11-13 duplication, 6 years old, 4 years since diagnosis)
3	'At the time [of diagnosis] it was really hard for me to hear. I just wanted to know, 'well, if this is his test results and these blood test results, then why can't you say he'll be delayed more in speech, or definitely have a disability, but he'll be able to maybe do X, Y, and Z, but gross motor might be more difficult for him or fine motor or whatever. That would be amazing if it could be more refined and more definitive of what those test results actually mean.' Parent 027 (two children with fragile X syndrome, 11 years old and 6 years old, 5 years since diagnosis)
4	'They had a register of families around the world who have people in their families with Angelman syndrome. I emailed them and I said, 'My son has Angelman syndrome, he has this really large deletion. I'm wanting to know if this would cause him to be more severe, is there any way that you can tell me about that?' … they gave me the contact details of five other families whose children had Angelman syndrome and according to the genetic information they supplied, had a similar size deletion.' Parent 023 (male child with Angelman syndrome, 3 years old, 2 years since diagnosis)
5	ET: How would you feel if the [prognostic genetic] test was not completely accurate? Would you still be interested? 'Well, I guess with any test, you're not going to know how accurate anything is. Like you can't say 'it's, you know, a hundred percent accurate.' You need to start somewhere. So, yeah, I would still be interested in doing the test. I just wouldn't take it for gospel.' Parent 004 (female child with 22q11.2 deletion, 7 months old, 6 months since diagnosis)
6	ET: If the [prognostic genetic] information was inaccurate, can you talk a little bit more about the potential implications of that? 'It's really same as a regular [unaffected] child. You can have someone who's got a very high IQ, but environmental factors will play a factor in whether they actually do achieve. […] It's not just the genetics, the tests which may or may not be correct, plus [there is] a huge factor of environment.' Parent 030 (male child with fragile X syndrome, 27 years old, 26 years since diagnosis)
Subtheme: Parents' acceptance of their child's condition helps acknowledge the irreducibility of uncertainty	
7	'I felt like there was something important about accepting that, sort of almost that kind of unconditional acceptance of a child and realizing that, diagnosis or not, no one knows [the future] and this idea of someone giving birth to a perfect child. It's fairly kind of problematic.' Parent 001 (female child with mosaic Down syndrome, 5 years old, 3 years since diagnosis)
8	'From Facebook groups and support groups, I've seen a lot of parents. A lot are in denial as well, so I think that the information will be more beneficial for ones that are in denial to help them accept and understand a bit better.' Parent 028 (two children with fragile X syndrome, 20 and 10 years old, 17 years and 8 years since diagnosis respectively)
9	'So, for me, I think you've got to get to that level of acceptance before you can actually process that [prognostic] information.' Parent 020 (male child with Angelman syndrome, 17 years old, 13 years since diagnosis)

sometimes traumatic. Receiving more precise results from a prognostic genetic test was forecast to be challenging, difficult to hear, and accompanied by negative anticipated emotions (Table 4, quote 10).

Parents thought they would need time to adjust to their child's diagnosis before considering receiving more precise prognostic information and suggested a two-stage approach for discussing or offering prognostic testing and information. A preference was expressed by parents for conversations about prognostic information to be positively framed where possible, discussing the child's potential strengths while also being realistic about their future abilities (Table 4, quote 11).

Parents discussed the potential for prognostic information to contribute to a loss of hope about their child's future, suggesting that psychological support should be available, particularly for those receiving less favourable prognostic information. Parents also discussed the potential impact of prognostic information on their child's sense of self and the possibility to cause harm (Table 4, quote 12).

Theme 3: Decision-making and motivations to receive prognostic information (parents are enthusiastic if the information will help with practical and emotional coping, although they stressed the importance of informed choice about prognostic testing)

Motivations for learning more precise prognostic information were described, despite the difficulty parents predicted experiencing when talking about their child's future abilities. Parents predicted that prognostic information would help with practical and emotional coping. Practical coping opportunities

TABLE 4 Illustrative quotes: Theme 2: Psychological needs and coping responses (conversations and decisions about prognostic information are emotional for parents, indicating the importance of psychological support).

In-text quote number	Verbatim quote
10	'If you had a child that really wasn't doing that great and they suspect his outcomes wouldn't be great, that would be really hard to hear at that point of diagnosis when you haven't had time to fully process it and work through your grief.' Parent 007 (male child with 15q11-13 duplication, 6 years old, 4 years since diagnosis)
11	'I think it just needs to be framed … if you're going to be making this sort of information available, it'd be good to also make available about the positive sides of things as well. So, present a picture that is more well balanced and not just talk in terms of risks or downsides or, you know, language that's negative or scary, but, you know, just sort of frame it in a more balanced way.' Parent 013 (male child with Phelan–McDermid syndrome, 5 years 6 months old, 3 years since diagnosis)
12	ET: Can you talk a little bit more about that, how you think people could respond to hearing about this sort of information about themselves? 'So, they might think they're an unworthy person to be part of society and might try and harm themselves or suicide, or might think they're a burden on their parents or their caregivers. You know, depression, anxiety that [learning prognostic information] could exacerbate those types of things, which are already prominent in this population.' Parent 015 (male child with 22q11.2 deletion, 12 years old, 2 years since diagnosis)

centred around the possibility that prognostic information would direct and prioritize interventions for their child as well as organize and plan for their child's future, including schooling decisions. Parents talked about sharing prognostic information with their child's care team, including allied health professionals and teachers (Table 5, quotes 13–15).

Parents thought that prognostic information could help them mentally prepare for their child's future and set expectations. In the case of more favourable prognostic information, parents predicted they would have peace of mind and the information would help to rule out the worst-case scenario (Table 5, quote 16).

Some parents made a distinction between motivations to receive prognostic information pertaining to physical challenges as opposed to cognitive and behavioural challenges (Table 5, quote 17).

Subtheme: The decision-making process about undergoing prognostic genetic testing (a risk–benefit analysis)

The decision to opt for (or against) more testing was predicted to be an easy decision to make, with limited decisional conflict anticipated (Table 5, quote 18). However, parents acknowledged the likely diversity of views on this topic, with the possibility for their co-parent to hold discrepant preferences. Parents described the importance of having the opportunity to discuss the decision for further testing with their co-parent and to make a joint decision (Table 5, quote 19). If the child was developmentally capable, parents planned to involve them in the decision-making process (Table 5, quote 20).

Parents anticipated they would weigh potential benefits and harms when making the decision to receive more prognostic information. The need for accurate information from a trusted source would enable engagement in this risk–benefit analysis. Trusted sources included the health professional primarily responsible for their child's care, experts in genetic testing technology, and support groups, including parents whose child had undergone testing. Most importantly, parents expressed the need to understand the possible test outcomes and how the test results would be used (Table 5, quote 21).

Theme 4: Parents' acknowledgement of possible societal harms of more precise prognostic information

Parents discussed potential societal-level harms with the availability of more prognostic information and categorization of individuals (Table 6, quote 22). While parents were overall supportive of genetic testing for more precise prognostic information and enthusiastic about improving the scientific understanding of neurodevelopmental conditions, they shared their fears about potentially unintended consequences. Parents questioned why neurodiverse individuals should be tested to determine their future IQ when neurotypical children would not be undergoing such testing (Table 6, quote 23).

Concerns were raised about prognostic genetic test results being used in pregnancy to inform decisions about termination (Table 6, quote 24).

There was a sentiment expressed that further testing for prognostic information should be offered as a choice to parents, and adequate counselling before testing and support for parents making the decision about further testing are essential (Table 6, quotes 25 and 26).

DISCUSSION

Our main finding was that while parents were generally enthusiastic about prognostic genetic tests, and their future implementation, they were more hesitant about learning such information about their own child. We found several different factors contributing to parents' preferences for prognostic information at both the individual and societal level. Some parents voiced concerns about the implications of this information for their child and family, such as the removal of hope. Societal-level concerns about the availability of such information contributing to stigmatization were also raised. Our findings are new in that previous research in other contexts found that parents have a desire

TABLE 5 Illustrative quotes: Theme 3: Decision-making and motivations to receive prognostic information (parents are enthusiastic if the information will help with practical and emotional coping, although they stressed the importance of informed choice about prognostic testing).

In-text quote number	Verbatim quote
13	'If that [prognostic information] can be used to target interventions and therapies specifically to what his needs are going to be like, then there's a benefit.' Parent 007 (male child with 15q11-13 duplication, 6 years old, 4 years since diagnosis)
14	'It's all about planning her services and getting her to the best place. For example, currently we don't live in a major city, but I take her to the city centre three times a week to special preschool. I don't know yet if she can actually attend the local school as I need to wait for her final year of preschool to go to the local school to see how they can support her and then decide if [they have] suitable supports.' Parent 003 (female child with 22q11.2 deletion, 4 years old, 4 years since diagnosis)
15	'And I think that [prognostic information] would help the therapist as well. It would focus their therapies and their expectations.' Parent 027 (two children with fragile X syndrome, 11 and 6 years old, 5 years since diagnosis)
16	'We went 12 months thinking the worst because we just thought a deletion was a deletion, we didn't really know that there were different aspects of it that meant, you know, you could be mildly impacted or severely impacted. So if they can give you not only the diagnosis, but then more information about the severity of that deletion, I think would have a massive impact on families and their ability to look at the future and see what that might hold for them and their child as well.' Parent 021 (female child with Phelan–McDermid syndrome, 7 years old, 2 years since diagnosis)
17	ET: You're focusing on getting more [prognostic] information around his more physical [challenges], but what about getting more [prognostic] information about the learning, cognitive, behavioural parts of his condition? 'That side of it doesn't specifically worry me as much. Yes, he's going to be delayed and his speech is quite delayed and you can't fully understand sometimes what he's saying, but that's not physically gonna hurt him. It's not going to kill him. He's not going to, you know, fall over and hurt himself because he can't speak properly.' Parent 011 (male child with 16p11.2 deletion, 11 years old, 9 years since diagnosis)
Subtheme: The decision-making process about undergoing prognostic genetic testing (a risk–benefit analysis)	
18	'Oh, then you just do it, like if there's no [physical] risk to the child, then I don't think I would deliberate much at all about whether to get that information or not.' Parent 008 (female child with rare deletion/duplication syndrome, 7 years 6 months old, 7 years since diagnosis)
19	'Weigh up I guess the potential of what that [prognostic results] could give us. My partner and I communicate really well about [child's name]'s diagnosis and what we want for her. We've discussed if there's potential for anything in the future that can bring any benefit to not just us, but future parents going through this, that we would discuss it heavily and make an informed decision. But we're pretty on board, with anything that comes available as long as it's not going to cause [child's name] any harm, obviously.' Parent 019 (female child with Angelman syndrome, 2 years old, 1 year since diagnosis)
20	'I would probably talk to him about it. He's 12 now. I might compare it to something like NAPLAN (National Assessment Program-Literacy and Numeracy) to put something concrete for him to compare with.' Parent 015 (male child with 22q11.2 deletion, 12 years old, 2 years since diagnosis)
21	'So first of all, I think we'd start with what the test does. What kind of information are we expecting? What's the accuracy rate?' Parent 016 (female child with Angelman syndrome, 2 years old, 1 year since diagnosis)

TABLE 6 Illustrative quotes: Theme 4: Parents' acknowledgement of possible societal harms of more precise prognostic information.

In-text quote number	Verbatim quote
22	'So, if we're putting kids into boxes of mild, moderate, or severe, are we not putting a label on what they can achieve in life? Is that not subject to a lot of prejudice and low expectations and things like that?' Parent 006 (female child with Sotos syndrome, 5 years old, 3 years since diagnosis)
23	'You wouldn't do that, you wouldn't go testing, typical chromosome counts and sort of say, 'well, you're in the average, and you're looking pretty fabulous. I don't know why you would perhaps be doing that … I feel like if it's something that would be ethically questionable about applying to the general population then [it is] worth giving extra consideration to if that's an okay thing for a particular group.' Parent 001 (female child with mosaic Down syndrome, 5 years old, 3 years since diagnosis)
24	'Well, if you give some parents that [prognostic] information while [they are] still pregnant, they may opt to abort.' Parent 003 (female child with 22q11.2 deletion, 4 years old, 4 years since diagnosis)
25	'I think you'd want to opt in, not just get it automatically.' Parent 020 (male child with Angelman syndrome, 17 years old, 13 years since diagnosis)
26	'I think if we are going to give parents that type of information, it's really important that they have the counselling involved as well, just to make sure that they are understanding it and focusing on the right side, you know, factual and supportive rather than emotional and stuff.' Parent 028 (two children with fragile X syndrome, 20 and 10 years old, 17 years and 8 years since diagnosis respectively)

to receive all prognostic information, have a low tolerance for uncertainty, and seldom consider societal-level implications.[25]

Parents reported that their tolerance of uncertainty evolved as their child aged, with some expressing a preference for prognostic uncertainty about their child's cognitive and behavioural abilities. The development of new technologies offering more precise prognostic information has the primary goal of reducing prognostic uncertainty.[3] However, our findings, along with other reports in the literature suggest that patients may embrace prognostic uncertainty and actively avoid prognostic information.[26] Our findings about prognostic uncertainty in the paediatric context offer a new contribution compared to previous research in the end-of-life context, where parents often will not have the opportunity to develop acceptance of uncertainty.

Our exploration of ambiguity as a source of uncertainty revealed that parents are generally comfortable with the potential inaccuracy of prognostic test results. Inaccuracies can arise when new technologies are introduced because of conflicting or limited scientific evidence. The concept 'ambiguity aversion' has been shown to impede decision-making in other areas of medicine, whereby those experiencing ambiguity avoid decision-making.[27] However, parents in our study were generally comfortable with the possibility for inaccurate medical test results, reflecting on their experiences with genetic testing, and this familiarity may counteract the effects of ambiguity aversion.

We found that parents prefer to receive prognostic information in a balanced manner, which includes both realistic and strength-based elements where possible. The way in which prognostic information is presented may impact how parents perceive and cope with uncertainty, and whether it is a source of fear or hope.[26] Future work could explore the impact of an optimistic versus pessimistic approach to framing prognostic information on several parent-reported outcomes. Similar work has been proposed in the neonatal intensive care unit, where parents nominate their preferred framing presented in hypothetical vignettes.[28]

Parents suggested that further testing for precise prognostic information should be offered as a choice. This suggestion raises questions about the most appropriate time for parents to be offered such a choice. Parents anticipated they would conduct a risk–benefit analysis when deciding about further prognostic testing. However, the emotions parents experience when discussing prognostic genetic testing may interfere with their decisions, causing parents to abandon highly analytical processes and default to using 'gut feelings' in decision-making.[29] Further work is needed to guide the implementation of the process and the timing of offering prognostic testing to parents.

Our participants generally supported the development of new tests and availability of more precise prognostic information. However, they also expressed worries about the potential harms of categorization, leading to further stigmatization. Such harms at the societal level are critical to consider when introducing new technologies. The process of categorization inevitably leads to the determination of a reference point, against which to compare different categories. Such a process intersects with the concept of ableism. Ableism refers to discrimination against people with disabilities and the belief that they are inferior to non-disabled individuals.[30, 31] Traditional biomedical models of practice have perpetuated ableism as a norm or 'default state'.[31, 32] Future work could investigate societal views more broadly to help understand the potential impacts of the availability of more precise prognostic information.

Finally, it is crucial to consider the context of the health care system in which prognostic genetic tests for neurodevelopmental conditions will be implemented. As genetic testing becomes more widely available because of increased government funding, more health professionals outside the genetics specialty (e.g. paediatricians) will be responsible for ordering and interpreting test results. Therefore, a comprehensive implementation science approach is necessary to ensure safe and effective clinical adoption of these tests.

Limitations

While our study provides valuable insights into the views and experiences of parents of children with a neurodevelopmental condition, it has limitations. Our participants were recruited from support groups; therefore, they may have a more proactive approach to their child's care and are motivated to receive detailed and accurate prognostic information. Additionally, the sample was limited in terms of ethnic and geographical diversity, with most participants being of Australian or New Zealand European descent. Our participants were more highly educated than the general population; however, there was representation across a range of educational levels. The time since receiving a diagnosis may have influenced parents' recollection.

Conclusion

We found that parents had mixed preferences and motivations for acquiring prognostic genetic information about their child. Our findings add to existing evidence that uncertainty can evoke both negative and positive emotions (specifically hope) in the new context of prognostic information for paediatric neurodevelopmental conditions. Health care providers should keep this in mind and consider presenting prognostic information in a way that highlights the potential strengths of the child when communicating test results to families.

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DATA AVAILABILITY STATEMENT

Data are not available due to data use agreements with participants and privacy.

ORCID

Erin Turbitt https://orcid.org/0000-0002-6650-9702
David J. Amor https://orcid.org/0000-0001-7191-8511

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SUPPORTING INFORMATION

The following additional material may be found online:
Appendix S1: Interview schedule.

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