

From Needs to Insights: Designing Digital Support with Parents of Children with Genetic Neurodevelopmental Conditions

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Abstract

Families find navigating a genetic diagnosis for their children with a neurodevelopmental condition complex and distressing, often facing a lack of tailored, strengths-based support. This paper presents the findings from a two-phase parent centred design study aimed at understanding parents' needs and exploring ideas to inform design concepts for a digital tool. Through interviews with nine parents, design workshops with ten parents, and collaboration of a multidisciplinary research team, our findings revealed a need for support that goes beyond clinical information. Key results highlighted the negative impact of deficit-based language, the essential role of peer networks for both emotional and practical guidance, and the invisible labour in care coordination. Our findings translate these needs into a vision for an empowering digital tool and offer a set of design implications focused on providing navigational clarity,

reducing care coordination burdens, and building parental agency by recognising the importance of designing with parents.

CCS Concepts

• **Human-centered computing**; • **Human computer interaction (HCI)**; • **HCI design and evaluation methods**; • **User studies**;

Keywords

Genetic Neurodevelopmental Conditions, Strengths-Based Approach, Human Centred Design, Digital Health

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1 Introduction

Neurodevelopmental conditions are chronic medical conditions that begin in childhood and affect how the brain develops, often leading to impact on learning, behaviour, communication, or intellectual abilities [31]. When parents or carers (parents hereafter) receive a genetic explanation (i.e., a genetic diagnosis) for their child's neurodevelopmental condition, they describe navigating uncertainty, conflicting emotions, and complex systems and information, both at the time of diagnosis and beyond [10, 33]. Parents report lacking tailored support and navigating inaccurate and negatively framed information emphasising their child's limitations [19, 22, 24, 38]. In addition, genetic support groups often lack sufficient funding to provide parents with the level of services and support they aspire to provide. Prior research to develop interventions that provide information and support to parents at this crucial time is limited, presenting an opportunity for a tool that can meet the needs of parents [17]. Such a tool may help parents to take a proactive and prepared approach by providing strength-based resources to support their child following a genetic diagnosis. A strengths-based approach is critical for reducing stigma and promoting inclusion by emphasising the unique abilities and potential of each child; for parents, this perspective reframes the focus from deficits to capabilities, enabling the development of more personalised, empowering and contextually relevant support strategies [12].

Previous studies highlight parents' desire for tools that are not only informative but also emotionally supportive, that include lived experience narratives, practical guidance, and condition-specific information [10, 33]. Our study aims to address these gaps by partnering directly with parents. Through this approach we aim to achieve two primary goals: (1) to understand parent experience and support needs relating to their child's genetic diagnosis, and (2) to explore initial ideas for a digital tool that can support parents in navigating their child's genetic condition. By acknowledging the lived expertise of parents of children with genetic neurodevelopmental conditions, our approach ensures the resulting insights are grounded in the real-world needs, preferences of families, and aligns with inclusive, strengths-based care principles [2, 4].

In this paper, we present findings from our two-phase study, outlining the challenges parents face, from navigating deficit-based language to the care coordination burden. This study is the first stage of a three-year consumer-led research project, NurtureNextGen, funded by the Medical Research Future Fund under grant number MRF2031116. The design concepts will be further refined through feedback and collaboration with parents as part of the project.

2 Related Work

This study builds on two primary research domains: digital tools designed to support parents, and design methods that involve families in the development process.

2.1 Digital tools for supporting parents and caregivers

Due to the rapid growth and accessibility of technology, researchers have explored digital support available to caregivers in varying contexts such as supporting chronic illness [32], mental illness [22], rare diseases [17], cancer [21], autism spectrum disorder (ASD), and

attention-deficit/hyperactivity disorder (ADHD) in young children [23]. Many digital tools support both individual and collaborative use. The most common features among these tools include web-based information and symptom tracking [17]. Some also facilitate information sharing [17, 19], self-reflection [32], social support [17, 19, 23, 32], emotional regulation [19, 32], and documentation of experiences and emotions [19, 21, 23, 32].

The ubiquity of mobile phones has led to mobile applications being considered in healthcare for their potential as convenient and effective educational resources, enhancing understanding of the condition and improving communication between parents and their children [17, 20]. Researchers have also explored the use of these apps to improve the quality of communication with medical professionals [17, 19, 23, 32]; for instance, Wu et al. [38] explored the quality of available digital information and how it can be used to improve communication with obstetricians for pregnant women during prenatal genetic testing. A number of significant limitations have been identified in existing tools, including incomplete or insufficiently comprehensive information; unreliable, outdated, or missing information sources; a lack of user-centred or evidence-based development; primarily deficit-based content; high reading level requirements; and the absence of visual materials, customisation options, or an effective text search function [14, 17, 38].

As genetic technologies advance and Medicare funding expands access, more families are receiving a genetic diagnosis for their child. For example, on average exome sequencing yields a diagnosis in 35% of cases involving neurodevelopmental conditions [25]. To our knowledge, few holistic digital tools have been designed that draw on the lived experiences of parents to positively support them during their child's diagnosis of a genetic neurodevelopmental condition. This gap motivates the exploration of a digital tool to provide strengths-based prognostic information to empower and support parents of children with genetic neurodevelopmental conditions.

2.2 Parent-centred design approach

Our study deliberately centred on parents as the primary participants. Parents bring deep insight and resilience to the evolving process of supporting their child after a genetic diagnosis. Consequently, the cognitive and emotional demands placed on parents after diagnosis are significant, including understanding their child's condition, coordinating care, and navigating complex health-care systems and disability services and supports, including the National Disability Insurance Scheme. Recognising these multifaceted needs and their lived experience, we engaged parents directly as experts in their own experience, to co-design a tool tailored to the needs and priorities of the intended end-users. This study adopted a participatory design approach, which aligns with co-design principles commonly used in health and social research. Both terms emphasise collaboration, but participatory design is used here to reflect the disciplinary context of human computer interaction. We acknowledge that participatory design can be time-intensive and demanding for parents already balancing significant caregiving responsibilities, yet we considered that the benefits of centring parent expertise outweighed these limitations.

Participatory design is a well-established approach that emphasises the active involvement of end-users throughout the design process [27]. The approach is particularly suited for research with children with genetic conditions and their families, as it promotes democratic engagement and ensures that resulting technologies are grounded in the authentic needs of those they aim to serve [18]. Participatory design involving families of children with disabilities often includes parents as key participants. This approach has guided the cocreation of social-robot behaviours for child mental health support [28] and the development of community interventions in Malawi that foster parental engagement in education [26]. A tele-practice redesign case study detailed a “how to” roadmap: it foregrounds relationship building, ensures accessibility, and balances aspirational ideas with service delivery constraints by working alongside a team of lived experience experts and staff [5].

Although strength-based participatory design has rarely been applied specifically to support and information needs relating to childhood genetic neurodevelopmental diagnoses, recent work underscores its promise. One study codeveloped online workshops that introduced parents of children with neurodevelopmental disorders to strength focused perspectives on health and development, with parents serving as collaborators and investigators throughout [29]. Another project iteratively redesigned a virtual therapy program, engaging parents and clinicians to align the platform’s content, language, and accessibility with user needs [11].

3 Study Design

Our study approved by the Royal Children’s Hospital Human Research Ethics Committee (HREC69604.2) and ratified by the University of Technology Sydney Human Research Ethics Committee (ETH22-7796). Our research followed a two-phase design. We began by conducting qualitative interviews with nine parents of children with genetic neurodevelopmental disorders to gain rich insights into their unique needs. In the second phase, we moved from understanding the problem space to co-creating solutions. This was achieved through two workshops: a workshop with the research team to frame possibilities, followed by a workshop with parents focused on generating concrete, potential solutions.

3.1 Phase 1: Qualitative Interviews

Participant recruitment: Participants were recruited using a purposive sampling strategy to ensure a diversity of experiences related to raising a child with a genetic neurodevelopmental condition. Recruitment occurred through partnerships with national and state-based genetic condition support organisations. The inclusion criterion was being a parent of a child with a diagnosed genetic neurodevelopmental condition. Nine parents participated in the study. Three parents had children with Phelan-McDermid syndrome, three with Angelman syndrome, one with Fragile X syndrome (more than one child affected), one with Mosaic Down syndrome and one with 16p Deletion syndrome (participant characteristics are displayed in Appendices A.1).

Format: Individual, semi-structured interviews were conducted over Zoom with each parent participant. These interviews were based on an Appreciative Inquiry model that focuses on improving through change and innovation, using four main topics: what is



Figure 1: Research team workshop

working well and why; envisioning what could be; how can this be implemented; and embedding this information into a sustainable plan [6]. Questions allowed for the interview to be adapted to the individual story. Each interview was audio-recorded and on average 35 minutes (ranging from 20 – 48 minutes).

3.2 Phase 2: Design Workshops

In the second study phase, we conducted two design workshops. The objective was to translate the exploratory needs identified in Phase 1 into more concrete design concepts and functional ideas for the digital tool. This phase comprised two workshops: an internal research team workshop and a subsequent parent workshop.

Participants and settings: The first workshop involved six core members of the multidisciplinary research team (Figure 1), including a consumer researcher who is the parent of a child with a genetic neurodevelopmental condition, whose academic and professional backgrounds include human-computer interaction, social science, genetic counselling, psychology, special education. All members possessed a minimum of three years of professional experience, with over half of the team bringing more than a decade of experience in their respective fields. The parent workshop included ten members of the study Consumer Advisory Group (CAG). All were parents of children with genetic neurodevelopmental conditions residing in Australia. CAG members brought diverse professional experience together with lived experience related to their child’s diagnosis and broader professional and voluntary advocacy involvement. This ensured a broad representation of lived experiences, with parents whose children have a variety of genetic neurodevelopmental conditions.

Workshop Format: The 90-minute format for both workshops was adapted from the 6-3-5 Brainwriting method [30]. Each session was structured around five main activities designed to elicit and refine ideas:

1. Introduction (10 mins): Participants were given time to review the core problem statement and key insights gathered from the parent interviews.

2. Warm-up Brainstorming (10 mins): To begin the ideation process, participants were asked to imagine key functions or challenges of the tool.

3. **Main Brainwriting Activity (40 mins):** In this core activity, participants engaged in six seven-minute rounds. They started by adding three ideas to a Miro board and then iteratively built upon each other's concepts in each round.

4. **Clustering & Discussion (20 mins):** Upon completion, all ideas were displayed and collaboratively sorted into thematic groups based on their conceptual connections.

5. **Voting (10 mins):** Finally, participants used three votes each to prioritise the most valuable ideas.

3.3 Data Collection and Analysis:

All materials, including audio recordings, interview transcriptions and the Miro board result were analysed independently by two researchers using reflexive thematic analysis following Braun and Clarke's method [7], with an open and iterative coding method. The codes were collaboratively reviewed and refined to ensure both depth and breadth in capturing patterns of meaning. The lead research assistant then finalised the thematic descriptions based on the agreed structure.

4 Results

4.1 Parent Interviews

The analysis revealed that parents navigate a complex and emotional path following their child's diagnosis. Our findings are structured around three key themes: (1) the choice and delivery of language; (2) the value of learning from lived experience; and (3) the call for actionable tools. Below, we present each theme with selected participant quotes.

4.1.1 The choice and delivery of language. Parents reported a clear contrast between two types of professional encounters. Interactions they recalled as negative, often with paediatricians or geneticists at the point of diagnosis, in which deficit-based language that focused on a child's limitations was used. P9 described this as being handed *"a list of your-child-will-never-do-these-things,"* an experience she found *"really confronting."* P4 concluded this approach was *"very much deficit based and negative"* and left her feeling *"helpless and hopeless,"* illustrating how negatively framed information can disempower parents, undermining their confidence and sense of agency. P7 expressed that the *"medical profession just needs to change the language when they're delivering diagnoses"*.

In contrast, positive interactions were empathetic, collaborative, and focused on the child's potential. These encounters treated parents as capable partners and provided tangible hope. An example came from a geneticist who, instead of making predictions, told parents their child *"will be the best guide to his own development."* P5 found this provided a practical and gentle framework, explaining, *"It wasn't just fluffy and like, 'Oh, anything can happen.' It was a really meaningful thing to say."* These positive encounters provide more than just clinical information; they offer parents tangible hope and reinforce their role as knowledgeable and capable partners in their child's care.

4.1.2 The value of learning from lived experience. Parents described the experience following a diagnosis as very *isolating* and highlighted peer networks and learning from those with lived experience

as a primary source of support. These communities provide emotional validation through a shared understanding that is hard to find elsewhere. As P2 explained, *"You just know. You just don't have to go through the pleasantries and small talk. You just kind of get it."* P3 found listening to other parents' stories *"so cathartic... knowing other people were going through the same thing."* Beyond this crucial emotional support, these networks also serve as a key source of practical, real-world information often unavailable from clinicians. For instance, P8 found recommendations for *"harnesses for the car"* and adapted clothing options through a private Facebook group. This advice is highly valued because it is grounded in the lived experience of others navigating the same daily challenges.

4.1.3 The call for actionable tools. Parents expressed a clear need for well-designed tools to empower them as advocates and in caring for their child and navigating the wider network of services and supports. There was widespread support for a question list to help them prepare for and get the most out of healthcare appointments, with P1 and P6 noting it would have been *"great to have"* during the *"pretty overwhelming"* initial diagnosis period. P4 explained it would have reduced the *"mental load"* of preparing for appointments and helped overcome the common feeling of *"you don't know what you don't know"*. Such tools are crucial for building the advocacy skills needed to assert, *"I don't want to be treated like I don't know what I'm talking about"*. P4 also identified a significant need for tools to manage a fragmented care team, describing the burden of *"passing on the information from this session to that session to that therapist"* and the frustration of *"starting from scratch each time"* with a new provider. These findings point to a clear opportunity to alleviate the significant and often unseen work parents do to manage and coordinate their child's care.

4.2 Design Workshops

The two design workshops generated 196 unique ideas (Appendices A.2). Of these, 95 were contributed by the research team, drawing on design and systems knowledge and lived experience. The remaining 101 ideas were generated within the parent workshop, reflecting responses shaped by everyday parenting experiences and practical needs. All quotes in this section were written notes by parents during the workshop

4.2.1 Research team workshop. Four major themes were identified from the research team workshop: a multimodal platform for shared lived experience, AI-driven coaching and communication, communication aids for self-advocacy and integration of digital and physical experiences.

A Multimodal Platform for Shared Lived Experience: A primary theme was the need to create a sense of community through shared experiences. Participants envisioned a platform that moves beyond simple chat functions, which are common in existing tools. Instead, they proposed a moderated space where parents can share and consume stories in various formats, including short, anonymous text posts, longer blog-style entries, and co-created audio podcasts. However, such a platform would require dedicated and expert resourcing for its moderation to be effective and safe. This oversight is crucial to ensure users do not inadvertently request or share medical advice, and to manage subjective feedback about

specific health professionals, thereby protecting both families and clinicians. This approach aims to provide comfort and solidarity while respecting user privacy and avoiding the pressures of direct social media interaction. Crucially, the platform must elevate lived experience as a vital source of knowledge which is valued alongside clinical expertise.

AI-Driven Coaching and Communication: There was interest in leveraging AI to provide personalised guidance and emotional support. Ideas included a custom-built virtual coach to help parents understand medical terminology, prepare for difficult conversations, and practice communication strategies. A key proposed function for AI was to help reframe deficit-focused language into positive, strengths-based wording. However, the team also acknowledged the significant ethical responsibilities, noting that any AI-driven feature must be carefully designed and verified by experts to ensure the information is accurate, trustworthy, and safe.

Communication Aids for Self-Advocacy: Participants identified opportunities for tools that support parents as they understand and advocate for their child's needs. The central idea was to create a *"communication aid"* or interactive database of questions that parents might ask clinicians and other health professionals such as an allied health professionals. This feature would not only provide a list of questions but also explain how to ask effective questions and allow parents to workshop and personalise them. The goal would be to provide parents with tools and resources that build on their existing strengths, supporting their confidence to navigate complex conversations ensuring their questions and priorities are understood.

Integration of Digital and Physical Experiences: The workshop produced many ideas that bridged the digital and physical worlds, recognising that parents may want to limit screen time. Suggestions included a physical *"tool kit"* containing items like a board game for the whole family, reflection cards, and stickers with QR codes that link back to the digital tool. This *"multi-sensory, multi-channel"* approach acknowledges that a purely digital solution may not meet the varied needs of families, suggesting the value of tangible artifacts that can be integrated into daily life. These physical components can facilitate family-wide conversations and activities that help all members, including siblings, understand the family's shared experience.

4.2.2 Parent workshop. The design workshop with parents produced a rich and detailed set of ideas, which were categorised into four primary themes. These findings articulate a clear need for a digital tool that serves not merely as a simple information repository but also offers meaningful support and guidance to families following a genetic diagnosis.

Navigational Clarity and Actionable Guidance: Similar to the results from the first phase study, the most urgent need expressed by parents was for clear, actionable guidance to navigate the complex systems they face after a diagnosis. Participants overwhelmingly voted for the creation of a *"pathway that shows exactly what should happen,"* including who to see, what to ask, and how to escalate concerns if support is not provided. This was conceptualised not as a static document but as a *"digital branching logic type tool"* that adapts to the users at different stages of adjustment and care. This navigational support needs to be localised, providing

suggestions for doctors, therapists and community connections that are local and have relevant experience.

Empowerment Through Communication and Advocacy: Parents desired tools that would support them as they advocated for their children. A central idea, which received strong support, was a *"self-advocacy tool/resource/question builder"* to help families prepare for ongoing discussions about prognosis. This would include prompts for strengths-based language to use with clinicians and family. The highest-voted idea in the workshop was for *"videos and practical advice on how to engage with your child/family (Including Dads)"*. These suggestions highlight a desire for a tool that respects parents' expertise and supports whole family engagement.

Peer Connection and Emotional Wellbeing Support: The value of peer support was a recurring theme, with parents emphasising the need for flexible and safe ways to connect. They proposed a system that caters to different personalities and cultural needs and people with low literacy levels, allowing users to connect in ways they are comfortable with, rather than through rigid, one-size-fits-all programs. Ideas included building a *"registry of willing peer mentors"* to connect with newly diagnosed families and creating safe, private spaces where parents feel they can share anything without judgment.

Integrated Technology to Reduce Cognitive Burden: Parents identified a major pain point in the invisible labour of care coordination and information management. There was a clear call for *"seamless communication of information between all healthcare providers and family"* to solve this. They pointed to existing concepts like the *"Rare Patient Passport [39]"* as a model for a platform where information can be shared at the family's discretion. In an ideal scenario, such a tool would interact with medical records to produce personalised reports on available government supports and even assist with applications for support and funding, thus significantly reducing the administrative and cognitive load on families.

5 Discussion

This section discusses the findings from our two-phase study. We begin by analysing the complementary perspectives of the research team and parent participants that emerged from the workshops, then we connect our findings to theoretical conversations within HCI and health. Parents' needs are framed through three conceptual lenses: reframing the narrative, navigating invisible care labour, and scaffolding self-advocacy. We conclude with a reflection on our methodology.

From Lived Experience to Design Imperatives: Our study revealed a gap between the clinical moment of diagnosis and the ongoing lived experience of raising a child with a genetic neurodevelopmental condition. Interviews exposed layers of emotional distress, isolation, and systemic fragmentation that far exceed a lack of information. This problem space shaped the design workshops, where our participatory, multidisciplinary approach became valuable. The parent collaborators, primarily focused on *what*, the tangible outcomes and practical functions the tool should deliver. The research team concentrated on *how*, the socio-technical systems required to enable these functions. This dynamic, further enriched by the inclusion of a parent-researcher on our team, created a

powerful synergy. The articulated needs of the parents guided the project's design, ensuring that potential solutions remained grounded in real-world problems and preventing a drift towards technology for technology's sake [34]. Our process, therefore, facilitated a productive dialogue between lived experience and diverse research perspectives.

Reframing the Narrative: Strengths-Based Design. Parents rejected deficit-based language and clinical framing that emphasised limitations. Their experiences echo critiques of the medical model that define conditions such as autism around impairments rather than strengths [13]. In contrast, parents responded positively to interactions that treated them as capable partners and focused on their child's potential. In contrast, positive encounters centred on potential and partnership were described as empowering. These findings align with the growing emphasis on strengths-based approaches and asset-based design in HCI [37], which emphasises the inherent strengths, skills, and assets that individuals and communities bring to the design process.

For a digital tool, this is more than a content consideration; it is a *design principle*. Information should be structured around goals and growth. Interaction design should support and extend parental capabilities, and the interface language must be affirming. However, such a tool will operate within systems that still use deficit-based paradigms. A parent might use the tool to frame their child's progress in terms of developing capabilities, only to enter a clinical appointment where the discourse reverts to limitations. This reveals a deeper challenge: the tool cannot simply be a repository of positive content. It needs to function as a counter-narrative and an agent of change, equipping the parent to navigate the dissonance between these two worldviews. The tool must empower parents to become translators and advocates, thereby giving them greater control over their family's narrative.

Making the Invisible Care Labour Visible. Where children with genetic neurodevelopmental conditions also have complex health and support needs, parents report significant time engaged in care coordination. Research shows that much of this coordination falls on parents as invisible labour [9]. Therefore, the design goal should not be to simply make the work visible to the system, but to make the outcomes of that work more portable and actionable for the parent. Rather than tracking activities for reporting purposes, a more effective tool might synthesise this work. For instance, instead of logging research time, the tool could support the creation of a concise, well-referenced summary to share with clinicians. This approach respects parents' roles and shifts the design focus from documentation to amplification, enhancing the impact of their existing labour rather than formalising it for external consumption.

Designing for Self-Advocacy and Empowerment. The parents' call for actionable tools can be framed theoretically as a need for scaffolding [16]. This concept, originating in the learning sciences and increasingly applied in HCI, involves providing structured supports to help a user build skills, confidence, and autonomy, with the supports gradually fading as their competence grows. The request for a "*self-advocacy tool/resource/question builder*" aligns with the use of *Question Prompt Lists* [35], a proven scaffolding technique providing caregivers with a structured list of potential questions increases their active participation in clinical consultations. Similarly, the desire for a tool that provides a clear pathway

is a request for navigational scaffolding. This is conceptually allied with tools like the *WWW-roadmap* [1], which guides parents through a structured inquiry process ("*What do I want to know?*", "*Where can I find it?*", "*Who can help?*") to prepare for appointments.

However, scaffolding must be adaptive. A newly diagnosed parent might benefit from basic prompts, while a parent with more experience may require advanced tools to compare therapies or connect with mentors. Designing flexible scaffolds that evolve with the user is critical and such a system would be a truly empowering long-term partner, promoting mastery and a sense of competence [3].

The Practice of Parent-centred Design in a Sensitive Context. The concept of vulnerability is not a static label but is situational [15]. In our study, parents' vulnerability is layered: it is cognitive and communicative when faced with complex information in high-stress clinical encounters, and it is institutional when navigating hierarchical healthcare systems. A core ethical mandate of our research was to avoid reinforcing these vulnerabilities. There is an inherent power differential between researchers and participants in any study [8]. We sought to mitigate this by explicitly positioning parents as experts in their own experience. Conducting research in such sensitive settings requires a reflective and empathetic practice [36].

The success of parent-centred design in this context depends on the creation of a third space, a safe, liminal space that is neither purely clinical nor purely personal, where parents can share their expertise, and researchers can contribute as facilitators rather than authorities. The multidisciplinary nature of our team, particularly the inclusion of a parent-researcher, was vital in constructing and maintaining this space. Beyond informing the design of new tools, a collection of these lived experiences could also serve as a powerful feedback resource for the healthcare system, helping to challenge deficit-based language and foster more empathetic clinical practice.

Limitation: This study has several limitations. The insights are derived from a small and relatively homogeneous sample of parents, who were predominantly of Australian European ethnicity and resided in metropolitan areas. This limits the generalisability of our results and highlights a need for future research that engages a more diverse range of families, including fathers, as well as families from culturally diverse backgrounds and those in rural and remote settings.

6 Conclusion

Our study employed a two-phase design process to understand the needs of parents navigating their child's genetic neurodevelopmental diagnosis and to generate insights to inform design concepts for a digital tool to support these families. Our findings revealed that parents require more than just information, they need a strengths-based ecosystem of support. The core needs identified were for clear, actionable guidance to navigate complex systems, flexible and safe connections to people with lived experience, and practical tools that strengthen parents' confidence and for their children.

The insights generated through our interviews and workshops have provided a deeper understanding of parents' needs, together with specific design opportunities and priorities. In doing so, our paper contributes empirically by surfacing parents lived experiences

after a genetic diagnosis, methodologically by demonstrating how participatory design can be adapted in sensitive health contexts, and practically by outlining implications for strengths-based digital support tools. Future work will focus on translating these findings into design concepts ready for user testing by parents of children with genetic neurodevelopmental conditions ultimately enabling the development of a high-fidelity prototype refined through iterative design and evaluation with families. By continuing to partner with parents as experts in their lived experience, we can create technologies that genuinely address their needs and foster a more hopeful and supported journey.

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A APPENDICES

A.1 Participant Characteristics of The Interviews

Characteristic		n	%
Child's condition	Phelan-McDermid Syndrome	3	33.3
	Angelman Syndrome	3	33.3
	Mosaic Down Syndrome	1	11.1
	Fragile X Syndrome	1	11.1
	16p Deletion Syndrome	1	11.1
Location	Metropolitan	7	77.7
	Rural	2	22.2
State	New South Wales	4	44.4
	Queensland	2	22.2
	Victoria	2	22.2
	Tasmania	1	11.1
Highest Level of Education	University or tertiary education	6	66.6
	Technical or further education institution	2	22.2
	Secondary School (Year 11)	1	11.1
Ethnicity	Australian European	9	100

A.2 Ideas from The Design Workshop

A.2.1 *Parents Workshop*. 1. List of general and specialised support groups (E.g. ACD /SWAN)

2. Make this an online tool so that it can be easily updated as things change.

3. Rebuild of the RARE Portal (launching by September 2025) will have a rare disease directory with links to relevant Australian support groups - this is rare disease specific, but you can ask for support groups to be added via the RARE Portal contribute paper. Happy to provide more information on this work. <https://rareportal.org.au/a-z-rare-diseases/> <https://rarevoices.org.au/a-z-of-support-organisations/>

4. Include more general groups such as support for carers.

5. Make this local to your state and area.

6. List of government supports (E.g. Centrelink/Carer Gateway/NDIS) to cover all basis.

7. Make this online or via an app as there can be changes to these supports regularly

8. Please visit the RARE Portal these are all highlighted there. The site has been rebuilt to improve accessibility/navigation in line with extensive stakeholder feedback

9. Rather than family needing to navigate a list of supports, in an ideal world, tool would be able to access medical records of patient and produce a personalised report of what government supports they may be entitled to and assist with the process of application, as well as suggesting specific community supports.

10. Again make this local to your area/state

11. Videos and practical advice on how to engage with your child/family (Including DADS).

12. adding where you can get additional support if you are struggling to engage with your family and child. who else can help. signs to be aware of to escalate concerns

13. This has similarities with my idea 3 and David's idea 3. Love this! Perhaps include a living list of practical communications tools/methods designed to communicate with children with different needs. A video of a dad peer mentor sharing tips on how they manage this would be powerful.

14. Also incorporate engaging / communicating with partners given the stress placed on relationships having a child with a rare genetic disorder

15. Perhaps a tool for dads to help them to feel empowered and what they can do if they are not the primary caregiver to support the primary caregiver. Plus information when parents are not on the same page to avoid challenges there.

16. to create a pathway that shows exactly what should happen when you receive a genetic diagnosis. Who you should go to, what you should be asking and how to escalate things if you are not getting the support that the pathway shows.

17. Practical steps/guidance on the pathways - perhaps a digital branching logic type tool e.g. if you are at this stage in your journey - do this, if that was a barrier 'go here next' - not sure how to describe this!

18. Ideally the pathway interacts with medical record or is visible to health care providers (hospital and community) as well as family, so everyone has visibility / accountability
19. Suggestions of paediatricians locally or who have experience with your child's condition. OTs/STs/PTs, who are local and have availability or experience with your child's challenges
20. Pathway flowchart/tool for GPs, Paediatricians and specialist but a separate flowchart/tool for families/parents etc
21. To show what a best practice approach to receiving a genetic diagnosis looks like. An overview of what the process should look like in an ideal scenario
22. Love this idea - what to do and what not to do perhaps? I think this is needed for health professionals. Should include who should be in the room during discussion on prognosis - e.g. should social worker/mental health be present or available. Families should also be given time to prepare and decide who they want in the room, if possible/not an acute situation
23. This should be focusing on the child's strengths not just deficits, avoiding 100% prognosis and instead language framed around possible pathways, not definitive. We don't know what the future holds or medicine. Also knowing disability is common, over 20% of the population is important to know.
24. Support mechanisms for families to support each other. this can be an incredibly stressful for family units and helping families access relationship counselling or grief counselling
25. Build registry of willing peer mentors and connect interested families to these if at time of diagnosis.
26. Local disability family support groups to connect in with such as Belongsides or Plumtree in each state. Books to read eg: Special by Melanie Dimmit
27. Exercise to support goal setting - for child and family E.g. Pictability
28. Integrate in to tool, so don't have to use external tools like Pictability
29. Goal setting that can be revisited throughout your child's life so we can celebrate the inchstones and invest NDIS \$ in the right areas!
30. Consistency with using tools like Pictability
31. Self-advocacy tool/resource/question builder to support family in ongoing discussion on prognosis
32. Make sure it is visible to all healthcare providers so discussions and communication focus on these factors.
33. In terms of prognosis, help parents to accept the reality of today rather than future-tripping and balancing that with planning the next 6-12 months ahead
34. Support tool/resource with strengths-based language prompts for talking about your child's prognosis to clinicians'/family/ friends
35. Produce communication that could be sent to other family members or friends, so families don't have to develop it themselves
36. Share examples of positive role models in the community with disabilities to normalize disability and help families understand 'ableism' and how to move beyond this
37. Seamless communication of information between all healthcare providers and family
38. An online platform where healthcare providers load summary reports and you have access to them.
39. A platform that allows information to be shared at discretion of the family (E.g. diagnosis paperwork)- therefore no repeating of stories
40. I would welcome a tool that means I don't have to repeat my story to every practitioner we meet. The concept of the RARE Passport (CAM RARE 'This is me' RARE Patient passport is a great evaluated tool that can be modified to suit different settings). The issue with these is getting the buy-in of hospitals and health services. (UK are leaders in this) <https://rareportal.org.au/personalised-tools-suitable-for-rare-disease/>
41. Personalised AI bot that can answer questions and provide information
42. AI bot can be accessed via your phone/app and has parents' knowledge and can access local organisations and suggest them and also has 'Melanie Dimmit' from Special her knowledge to help parents feel positive about the journey and understood!
43. Just wondering what this would focus on and where it would pull data from? Important to recognise how different the Australian context is compared with other countries.
44. Generate age-appropriate communication to use with child being diagnosed
45. An AI bot that grows and adapts with your child
46. This is so needed. Parents often left to their own devices to decide how/what/when to communicate. This would need to cover off all abilities /types of communication
47. Include state-based service providers in the tool and state-based communities
48. Include interactive map to be able to choose services in your local area or be able to plan for moving house/interstate etc
49. See 'Services' tab on the RARE Portal. The new site will have interactive blocks for each state and territory. If there are any state specific resources already available that support prognosis discussions please let us know :) <https://rareportal.org.au/#>
50. Use AI to share hopeful advice from other parents who have been there
51. A searchable directory of hopeful advice/ personal stories would be nice! Could add to this 'best practice' stories and maybe even stories of what not to do. Could be a great place for training medical professionals to learn the dos and don'ts when interacting with patients, including during discussions about prognosis.
52. The Now and Next program is an exceptional one from Plumtree - Build on this, include NDIS set up tool.
53. Love Now and Next too! <https://www.plumtree.org.au/for-families-by-families/now-next/>

54. Links to supports services (both peer support and specialised health services)
55. Be aware of social media comments, take them in their stride. Don't feel pressured by other people comments.
56. There are so many that some kind of interactive tool to filter & find resources including research peer support and patient orgs
57. Yes, not feeling alone and isolated. For such a long time after diagnosis it feels like there is no one else. Having that connection from day 1 could help with mental health a lot.
58. Ability for researchers to see de-identified data with ability to reach out to patients as needed
59. Consent is an issue as patients with rare diseases can be identified as there may be very small numbers.
60. Perhaps links to existing data repositories to not reinvent wheel
61. Love this concept as a way to share lived experience. I'm sure the bugs could be worked out.
62. Links to a national database (e.g. Garvan Institute) of all people living with a genetic condition
63. What the controls around these registries.
64. & global for very rare
65. There are already some for ASD and Fragile X was included. Would be great to combine them all. There is so much cross over.
66. Talk about the benefits of peer support
67. think about different levels of support for different people - cultural, shy, ready to get in and get involved.. Perhaps a set of questions to determine best way to meet needs
68. Allow people to connect in ways that they are comfortable with. Don't put strict rules of how, when and for how long in place (I have a particular issue with what they did with MyTime as to why I say this).
69. Agree - definitely need flexible options to cater for all personalities/lifestyles
70. Encourage sharing information and resource
71. Build resource library categorised under need
72. It needs to be a safe place where people can share anything. Sometimes what we do as special needs parents is outside the 'norm' and we can't be judged for it.
73. Have some 'ground rules' when signing up/using the tool e.g. no sharing information, no screenshotting etc
74. Parents are their child's best expert, don't underestimate your gut instincts
75. Educate to professionals about this, parents should be a resource not a hinderance. Dr Harry Angelman was known to tell his students "An Angelman mum is your best resource, listen to her"
76. The lived experience of parents should be respected and should never be belittled. Just because something was learnt at university doesn't mean what the parent says is wrong or invalid.
77. Engage those with lived experience in more research in this space
78. Talk about connection & its value & it can be tailored for individuals need
79. Being able to connect with other families who have a similar diagnosis can be a safe space and help cope with grief and daily living.
80. Need contact details for organisations who facilitate this and the tool can assist with 'warm handovers'
81. Spot on, I always say "never underestimate the value of peer support"
82. Don't discount things that may happen later in life but help parents deal with the now
83. Concrete help on how to cope with the early years. E.g. if sleep issues are common with that diagnosis, what are tips and helpful ideas to help.
84. Also links to relevant specialists/therapists/peer support organisations operating in that space
85. Every child will tell their own story. We don't always know their trajectories of rare genetic conditions.
86. People get caught up in what it means for their child but they are still individuals with many awesome qualities. Ie. Your child has Angelman, they are not Angelman
87. Helping parents find ways to get support and respite. Sometimes getting away for a break can be and enormous help. Linking parents with counselling and emotional supports.
88. Provide listing of support organisations in each state that provide respite
89. Every child has the right to play and socialise, their disability should not stop them. At the end of the day, they are just kids.
90. A list of services and therapists with experience in the diagnosis you have just received.
91. Also encouragement to consumers/carers to access these services (e.g. videos/audio clips from those with lived experience saying how important these services are as part of the overall journey - such as peer support, mental health etc)
92. Be aware that services and supports change, so ensure the list is up to date. Not every therapist suits every child, there is nothing wrong with changing.
93. This again might be an interactive tool that professionals are encouraged to sign up to if they feel expert in certain conditions
94. A link to other parents and families who are living with the same diagnosis.
95. Also link in with organisations across Australia who provide umbrella peer support e.g. Peer Support, Disability organisations
96. Talk about the benefits of peer support.
97. this would need to be an opt in service, would you do separate to patient orgs? Is this a consent - I consent to be connected... Then you could potentially match people with a "buddy"

98. Information about research that has been done and is ongoing to help with symptoms and/or work on a cure. A way to continue to follow along with that research. The ability to put your name down to be part of research or test phases if you wish.

99. Some ability for two-way communication with the researchers, and also regular updates on new research and clinical trials e.g. get notifications

100. Provide opportunities to partner and participate in research opportunities.

101. This is important, you are often told "no treatment" but there may be a lot in the pipeline.

A.2.2 Research Team Workshop. 1. How does this avoid turning into a chat feature that is already available in other digital tools? What can this tool uniquely provide. 'Characters' brings me back to the idea of avatars and GenAI.

2. Short snippets of advice/information/stories provided daily in the style of an Instagram account, with the advantage of not having to use social media, sponsored content, advertising etc.

3. Providing support and comfort in solidarity and shared experiences - users do not have to interact with each other, though maybe there is a way that users can submit anonymous posts which are moderated and shared.

4. There could be the opposite whereby stories of grief, sadness, sitting with negative feelings are openly acknowledged and explored.

5. Great, for short personal information this could be similar to the Say Their Name tribute wall - where anonymous messages can be posted that will scroll across the screen for anyone to read: <https://www.saytheirname.org.au/your-tribute-wall> If it's longer messages, then the format could be in the shape of blog posts on websites.

6. Allow people to share their experiences that those feelings are okay and that they're not alone and give them opportunity to explore the possibilities together or share strategies that may have worked for them in the past.

7. Parent stories about how they adapted to receiving their child's diagnosis.

8. a tool that can share some real experience of parents. the focus will be on strengths.

9. invite parent to record their experience with "experts" and share with others

10. The real experiences are co-created with parents and families, presented in an audio format (e.g. a 10-minute story/podcast). There could be an audio element. The stories could be presented from the perspective of a child/person living with a genetic difference to offer parents a peak into a hopeful future.

11. a website to read other parent's real story, animated?

12. The tool can include a set of short podcasts, some for parents, some for whole families, to listen to - e.g. while in the car - that talk about relevant experiences of similar families. This can be both light-hearted as well as serious, a mix would be great. Parents can suggest what they would like to hear in these podcasts and maybe make suggestions in the tool for podcasts still to be recorded. This can be low-tech, e.g. Waffle Wednesday - I'm happy to explain!

13. Parent stories provided in a range of different formats, including a hand-out/newly diagnosed tool kit (physical), as well as digital, written, audio, video.

14. This could work evidence-based information and lived experience together - weave together. Could also challenge the notion of who is the 'expert' - not just clinicians as experts. Give voice to lived experience expertise.

15. Parents would be provided a range of prompts to share their experience, reflect on it and consider what they might do differently next time e.g. "I didn't like how I wanted to ask a question, but I forgot because I was overwhelmed, so next time I will write down the question and show it to the health professional". The tool could also provide advice based on the reflections of what parents could consider doing differently next time.

16. use post, or group chat (waterfall information), or create a chance for new parents to get to know existing group or community.

17. A virtual, regular meet-up for parents that is facilitated by a parent (peer to peer mentoring/support)

18. Connecting Parents in the community to share experiences, tools, guides that worked for them also asking for support

19. Online video meetings between parents can be a great opportunity to build a network, share concerns or ask questions that you would not want to post. This meeting could also be used for the children to meet each other

20. I worry that this online meet up option is available through the envisage program. I wonder about the opportunity to be matched with a peer mentor instead?

21. Filling a gap that other digital tools don't - e.g. not just a tool with chat functions

22. AI custom built Virtual coach to chat, ask questions, improve understanding of condition, get the general idea, to prepare for discussion with medical team etc. Also personalized and adapting to needs. Risks: need careful consideration

23. A step-by-step coach, explain medical terms.

24. The tool could show various ways in which parents can respond to issues, e.g. a child being bullied. It could give suggestions for how to talk to school, to the bully, to the parent of the bully, but also how to support the child in understanding how to deal with bullying, for example

25. AI can be a great way to 'translate' medical information to parents, but it would require an expert to verify the information and/or the AI needs to be trained specifically for this aim, to be trusted, and/or it should only write about topics it cannot really make mistakes about.

26. An AI tool that explains genetic reports to parents in patient-friendly language that is also strengths-based.

27. a personalized conversation agent can build different template for diverse users

28. AI-DRIVEN coach? to give you suggestions.

29. Talking about strengths is a difficult skill, explore with coaches
30. I think we might be able to use GenAI to automatically make language more strength-based and positive in its wording. The tool could suggest 3 alternative phrasings and let the parent/user improve the app by picking the best representation? Over time the info will get better!
31. If AI presents quality control issues could go with an audio/podcast of an expert and a parent perhaps talking through some of the trickier common words in this space
32. I wonder if it is less a mentor and more in the direction of buddy. Like when our kindy students are paired with an older buddy say in years 5 or 6.
33. There's also AI generated simulations now - develop the back end on evidence-based information, ensure user testing for various aspects including accuracy of information, tone and style of response.
34. A coach that parents can use to practice conversations with people in a range of settings, with examples from people with lived experience (e.g. "what should I do if my child is being bullied" - there would be a matching (AI?) parent response of someone who has navigated this with advice for what helped/didn't help. Mimicking an online community where you might ask e.g. a Facebook group but in a more controlled way.
35. Our tool could chat with our parents (either through text or audio) to help present the most useful information to the parents. This could be the tutorial when starting to use the tool to later customize the user interface and experience (customization as opposed to personalization)
36. Some examples of deficit focused wording used in a diagnosis/prognosis conversation and a 'fixed it' version that is strengths based
37. The tool could include a special functionality: preparation of a (difficult) conversation, reflection on how the conversation went
38. A communication tool that allows parent to view (fact-checked, peer-reviewed) information and sharing experience with others.
39. A tool that includes lectures or experts' advice (Q&A?) can be easily accessed. Aim for teach parent about how to ask the right questions.
40. FAQ's including where parents have gone to find answers (demonstrating many questions will not be best answered by clinicians)
41. Parents are provided with a list of questions they might find useful to ask their doctor to elicit strengths-based responses. Parents can also use the tool to role-play asking the questions and workshop the questions to personalize them.
42. The opportunity to explore, think carefully about the language used in questions and what underlying assumptions this might overt. Could have a table of 'what do I want to know' and a range of useful questions that might best elicit this (also important to understand clinician not best placed to answer all)
43. Experts could be asked for the best questions parents could ask. Parents could upload the questions they had asked or wished they'd asked and explain why they were/could be useful.
44. The database with potential questions to ask your doctor, could also include a short explanation on how to ask questions (open-ended for closed, e.g.), and remind the parents that they have the right to ask questions, to encourage them to ask anything they like
45. Parents could populate what questions came to mind for them. They could compare this to other parents' experiences and essentially create a lived experience question bank.
46. A database including example questions to help parents interact with health professionals, support workers, friends, family, teachers, anyone who may come into contact with the family (even responses/comments from strangers). To some extent helping parents and families advocate for their child (or themselves).
47. Tools/skills to help parents communicate and ask questions in sessions with health professionals.
48. a function that can allow parents to ask questions and invite experts or other parents to review it, provide feedback. The interface will save the process.
49. Interactive Questions/quizzes /prompts to provide guidance on type of questions to ask. Can a question bank be interactive where parents add but doesn't become overwhelming
50. tool book? a notebook? a database to include example questions or real experience to help parent how to interact with health care professionals
51. a collection starting with 'what do I want to know' and suggestions for the best questions/language to use based on what has worked well for other parents
52. This idea could be extended beyond written communication to images. Or could be crafted into other 'easy read' or alternate formats for digestion/distribution beyond inside the tool.
53. Meet the experts - a range of characters built on pooled lived experience who can respond to Q and A's
54. A card game to help parents and children find interest in common.
55. Activities to do with Children to understand their areas of interest etc. in different formats
56. This idea made me think of a GenAI based robot I've seen used to support mental health in children and children who are neurodiverse - its name is Moxie
57. A board game that the whole family can play (both parents, siblings etc. including extended family members through virtual connection, also friends) that helps the family, friends and extended family understand the experiences that parents and their child are going through and find common interests and topics of conversation.

58. Love the board game idea because it engages all ages into a physical activity, where the topic of everyday life could be talked about. An inspiration could be the board game that my PhD student Nandini developed to create gut awareness in people, which is also about everyday life and the consequences of actions

59. wonder about a teddy bear - think feel do bear - focused on adapting to new information

60. like a real game, bingo or ...? a corner just for parents, allow parents to express themselves.

61. The physical tool kit could include the boardgame, question and reflection cards, a colouring book, a diary, stickers with funny and supporting statements for the children to use, and stickers with QR codes that link to the digital tool, that can be stuck on anything as a reminder.

62. A type of mural board? Posting photo's, reflections, challenges and 'wins' (for lack of a better word).

63. meditation, and some activities that parent can do with their child and have the chance to express their love and recognized strengths.

64. This can incorporate a range of different activities according to the child's needs. E.g. it might be physical touch/massage, have a dance party, do some drawing or colouring with a particular prompt that encourages strength reflection.

65. a interactive photo frame that allow child to use to tool to draw on it. user can upload photos.

66. Tool can present (fictitious or real) stories (scenarios) of families living with a genetic disorder, showcasing the varied rewarding experiences they have had, but also issues they run into and beautiful workarounds/solutions that work well for them. These stories could be a pleasure to read and engage with, perhaps as bed-time reading for children with beautiful drawings, that parents and child can talk about.

67. 'Yes, let's' dramatic play/game. Homie with another Chromie merchandise too.

68. The fact-checked, peer-reviewed information would be framed in a positive, strengths-based way. For example, if the information said "children with this genetic condition are unlikely to ever talk" the tool would translate that to say "children with this genetic condition may use alternative ways to communicate other than verbal". With some short videos of people communicating effectively, positively and inclusively using that method.

69. A co-created prognosis document inclusive of strengths identified by parents (and not called a prognosis document!)

70. More interactive information available in bite sized information

71. Tool adapting to the needs, being able to provide the information that you have to adapt and give suggestions

72. The document could include: Developmental areas/Personality strengths/Motor/speech and social skills. How would this incorporate such individualism in prognosis?

73. This makes me think of noting that even getting the same diagnosis doesn't mean the same prognosis - unique journeys and often not matching an 'expected' prognosis journey or one originally portrayed/communicated at point of diagnosis to the parents.

74. Depending on the stage of life at the time of diagnosis allow the lived experience to inform and guide the existing available information

75. Explore tools on diversity and inclusion and how they support diverse people

76. A 'scene setting' opportunity to bring in concepts such as 'strengths based'

77. Could include an emotional support element - encouragement that parents are doing a good job

78. Giving opportunity to allow parents to sense check at multiple stages if it is still relevant? to ensure they get most suitable adaptation as per current needs than what may have worked in the past

79. Could include an emotional support element - encouragement that parents are doing a good job

80. Recognizing that there is an emotional toll on parents of children with disability who often expend additional energy navigating systems (the world!) which are not built for their child/family. 'I wouldn't change you for the world, but I would change the world for you'

81. Yes, recognizing the physical energy and mental toll of advocating for our littles. Elucidating/bringing that into light in a feature.

82. Could explore different layers of lived experience e.g. emotional processing, physical changes to home, work or other environments, cognitive/thinking/mental health.

83. There could be the opposite where by stories of grief, sadness, sitting with negative feelings are openly acknowledged and explored.

84. Drawing on parents strengths and likes, not just focusing on their child's strengths. In an ideal world, if you had time, what would you do for yourself that would bring joy?

85. Self-care could be a feature in the tool, that is both designed to support parents in their network, as suggests self-care activities for the child. As inspiration I suggest the app Finch (which I am happy to demonstrate).

86. Alternative formats can be excellent as provide opportunity for accessible information for people with different needs and thinking about parent's busy and overwhelming schedules

87. Can also incorporate photos or short videos that can be looked at/watched and also shared with other family members and friends. E.g. if a friend sends a text to a parent "how are you? how is your child?" parents can simply provide a link to the story/photo/video that updates the friend on their life. Parents can find it hard to respond to messages, it takes mental load. The default response is "my child is doing great!"

88. an interactive storybook, parent and child can co-design it, (like record their voice to make it interactive), use before bedtime.

89. A tool that can record parent and child's daily activities, combine with real-life game (card game, sport). When people review it, people will have positive emotion.

90. The tool should not just be serious, there should be a fun corner, e.g. with random stuff people can post or fun things to do with your family.

91. The record of daily activities could be submitted using photos. Parents upload a daily photo of an activity/memory/event. The photos are then made into a monthly video. If parents forget to do this daily, they can still upload photos at a later time, and the tool will find a photo with the corresponding time stamp.

92. Parents can upload media (photos or videos) whenever they feel like but are rewarded at the end of 25 uploads (or similar), either through a summary video or a physical photobook (as inspiration: Polarsteps app - I am happy to show what it looks like!). Parents can choose to share their media with trusted friends who are also members of the tool.

93. An opportunity to create a personal 'road less travelled' by uploading photos - prompts could be provided to support strengths-based approach

94. Type of Mural board may become quite difficult to manage overtime? Additional opportunity to organise memories and reflections overtime to be filtered based on key words and artificial intelligence

95. a managed mood board, or waterfall information, real-time updated, have achieve for past information