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ORIGINAL ARTICLE

Exploring the journey to genomic testing and genetic services: A qualitative study of parental perspectives of children with rare conditions

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Abstract

Despite affecting a small portion of the population, rare conditions have a significant impact, collectively affecting around 300 million people worldwide. Historically, early diagnosis has been impeded by failure to recognize rare conditions and order/refer for appropriate genomic testing. The advancements in genome sequencing offer a more agnostic and accelerated approach to the identification and diagnosis of rare disorders, potentially improving health outcomes, reducing the impact of disability, and reducing financial and psychological burdens on families. Due to the complex nature of these conditions, early engagement with genomic testing and clinical genetics services is key to facilitating a diagnosis. This qualitative exploration aimed to understand the journey to genomic testing and services and identify the supports families need during the diagnostic period. We conducted semi-structured interviews with 24 parents of children with a rare condition. Interviews were analyzed using inductive reflexive thematic analysis. Three themes of the parent experience were identified (1) the need for a streamlined pathway through the healthcare system, (2) the value of healthcare professionals who listen to parents, believed them, and partnered with them, and (3) the power of accurate diagnosis. Our findings indicate that providing direct and timely access to genomic testing for patients with a suspected rare condition could alleviate psychological and financial stressors. Genetic counselors are adept at supporting families affected by rare conditions and are optimally placed to facilitate timely access to genomic testing. Improving timely access may be facilitated through educating primary care physicians and embedding genetic counselors in pediatric settings.

KEYWORDS
diagnostic odyssey, genetics services, lived experience, parents, rare condition, rare disease, referral practice

1 | INTRODUCTION

Rare conditions are defined as those that occur in less than one in ten thousand individuals (Zurynski et al., 2008). There are over seven thousand known rare conditions, cumulatively affecting 6%–10% of the global population (Bhattacharya et al., 2021; Rare Voices, 2021; Schieppati et al., 2008). Most rare conditions manifest in childhood (Zurynski et al., 2008) and up to 80% are estimated to be caused by an underlying genetic susceptibility (Chong et al., 2015). Families of children with rare conditions

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often attend numerous medical appointments and undergo multiple tests before receiving a diagnosis (Anderson et al., 2013; Wong et al., 2015; Zurynski, Deverell, et al., 2017). The extended journey to achieve an accurate diagnosis is referred to as the "diagnostic odyssey." The duration of the diagnostic odyssey for rare conditions varies considerably between individual cases, and delays of up to 30 years have been described (Molster et al., 2016; Sawyer et al., 2016). Forty percent of patients with a rare condition are misdiagnosed at least once, adding to the time it takes to receive an accurate diagnosis (Schieppati et al., 2008; Zurynski, Deverell, et al., 2017).

Parents living without a diagnosis for their child's condition experience a variety of psychosocial impacts including increased levels of uncertainty, decreased capacity to cope, and a myriad of feelings leading to emotional vulnerability (Dellve et al., 2006; Graungaard & Skov, 2007; Madeo et al., 2012; Spillmann et al., 2017). Inadequate psychosocial support during the diagnostic period and the significant financial burden of rare conditions on families and health systems has also been reported (Anderson et al., 2013; Chung et al., 2023; Sequeira et al., 2021; Yang et al., 2022). Additionally, living without a diagnosis can limit access to financial, medical, and disability support (Anderson et al., 2013; Pelentsov et al., 2016; Soldatic et al., 2014; Yaneva-Deliverska, 2011).

Genomic sequencing can provide rapid, accurate, and cost-effective results for patients with rare conditions, potentially transforming the diagnostic process (Lunke et al., 2020; Stark & Scott, 2023). There is growing evidence internationally for the clinical utility of whole genome sequencing as a frontline test (McDonagh et al., 2021; Stark & Ellard, 2022; Turro et al., 2020). Recent studies of rapid genome sequencing in acute care settings have shown average diagnostic rates of 33%, with result turnaround as little as 3 days in critical care (Byrne et al., 2023; Lunke et al., 2020; Scheffer et al., 2023; Stark et al., 2023; Temple et al., 2022). However, there are major translational barriers affecting the use of these technologies in healthcare systems. Healthcare providers (HCPs) report limited knowledge and confidence in implementing genomic medicine (Mordaunt et al., 2023). Additional barriers to genomic medicine for frontline HCPs include complex result interpretation and counseling, lack of infrastructure, and lack of local experts (Biswas et al., 2020). These barriers lead to lower uptake of genetic services and contribute to diagnostic delays for patients with a rare condition (Anderson et al., 2013; Wong et al., 2015; Zurynski, Deverell, et al., 2017).

Providing integrated approaches to improve equity of access and treatment in rare conditions has emerged as a global priority as highlighted in the 2021 United Nations resolution, "Addressing the challenges of persons living with a rare disease and their families" (Halley et al., 2022; United Nations General Assembly, 2022). Common objectives across national rare condition initiatives are to achieve effective and accurate timely diagnosis, equitable access to the best available technology, and integration of mental health and social and emotional support for rare condition families (Bhattacharya et al., 2021; Chung et al., 2022; Halley et al., 2022). In support of these priorities, this study aims to better understand family perspectives on the challenges in obtaining a genetic diagnosis.

What is known about this topic?

A protracted diagnostic odyssey incurs significant psychological and financial strain on families. The diagnostic yield from whole genome and exome sequencing has accelerated their integration into primary testing pathways, particularly in neonatal and pediatric acute care settings. Internationally, rare condition initiatives emphasize the importance of equitable and timely access to genomic services.

What this paper adds to the topic?

This paper highlights that families require a more streamlined pathway to genomic testing and services to mitigate psychological and financial stress associated with a protracted diagnostic odyssey. Parents valued partnering with invested primary care providers and pediatricians who accelerated the diagnosis and validated parental concerns and expertise. Cumulatively, these findings support international momentum to implement genomic testing for patients with a suspected rare condition as a first-line test with adequate health system and health provider support.

2 | METHODS

This study was approved in Victoria through the Royal Children's Hospital in Melbourne, Victoria (HREC Approval: HREC/58138/RCHM-2019 RCH reference: HREC 2019.250) and in Queensland through the University of Queensland Human Research Ethics Committee (HREC) (UQ/AMO3561). The HREC approval for Queensland was further ratified and approved by the University of Technology Sydney HREC (UTS/ETH19-C0004).

2.1 | Study design and recruitment

This study used a qualitative study design. Eligible participants included parents of children with diagnosed or suspected rare genetic conditions in Queensland and Victoria, Australia. Participants were identified and recruited though advertisements on the closed Facebook page of the patient support group: Syndromes Without A Name (SWAN) Australia. A recruitment text was included in SWAN Australia's newsletter, website, and X (formally known as Twitter). All individuals who expressed interest were invited to participate.

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Informed consent was sought from all participations via a written participant information and consent form.

2.2 | Procedures

Semi-structured telephone interviews were conducted from December 2019 to August 2020 by authors (MK) or (CW). Semi-structured interviews were chosen to maintain focus on the journey to genetic testing and services while allowing researchers to delve deeper into participant insights and provide opportunity for clarification. At the time of interviews, female researchers MK and CW were both genetic counseling students with no children and little experience working with families with rare conditions. MK and CW conducted interviews separately in Victoria and Queensland, respectively. Questions explored the participants' experiences leading up to their referral to genetic services, experiences with different health professionals, facilitators, and barriers to clinical genetics referrals, and the impact of receiving a diagnosis for their child. The interview guide can be viewed in Appendix S1.

2.3 | Data analysis

All interviews were audio recorded, de-identified, and transcribed by MK and CW. Reflexive thematic analysis (RTA) was chosen to analyze the data to ensure an authentic representation of participant experiences and contextuality. The positionality statement and the Reflexive Thematic Analysis Reporting Guidelines (RTARG) responses, as recommended by Braun and Clarke (2019) can be seen in S2 and S3 respectively. Initial inductive RTA was conducted separately by MK and CW guided by Braun and Clarke (2019). Separate analysis occurred initially because MK and CW were operating remotely in separate states and the data from each state was used for Masters of Genetic Counseling dissertations at different institutions. Both researchers used a similar process of familiarization by listening to recordings and reading transcripts. MK and CW then individually assigned preliminary descriptive codes to each interview in an iterative manner to features that stood out about parent navigation to genetics. During this iterative process, authors MK and CW met regularly with author, supervisor, experienced genetic counselor, and qualitative researcher AML to discuss common experiences from the interviews and what ideas needed further exploration. Recruitment was suspended when MK, CW, and AML deemed that new interviews were replicating ideas from previous interviews (i.e., thematic saturation), and there were significant similarities between the experiences reported in separate states. Discussions between authors MK, CW, and AML were then undertaken to refine preliminary codes and create initial subthemes. AC and AML revisited these subthemes in the context of increasing availability of pediatric genomic testing in 2023. A constant comparison approach was used to reanalyze the entire dataset to identify prevalent themes for parents in both states. This was highly salient as publicly funded pediatric genetic testing had subsequently become available. This allowed for the discussions of the findings to be contextualized relative to the changing landscape of genetic testing for rare disorders. AC is female, a parent, genetic counselor, high school teacher, and of Australian First Nations ancestry.

3 | RESULTS

3.1 | Participant characteristics

Twenty-four parents from twenty-two families consented to participate from the SWAN closed Facebook page of 485 members. Most families had only one child affected by a rare genetic condition; however, there was a small number ($n=3$) of families with two or more children affected. Most participants (22/24) were female. The average duration of interviews was 40 min (range: 20–87 min). Twenty-two children had a genetic diagnosis, most commonly made via whole exome sequencing (15/22), followed by a chromosome microarray (7/22). Two children in this cohort remain undiagnosed (ID numbers not noted for confidentiality). Individuals experienced varying lengths of diagnostic odyssey, ranging from birth to 8 years. Five of the twenty-four children were misdiagnosed one or more times throughout their diagnostic journey. Broadly, conditions included bone growth disorders, intellectual and developmental impairment, hematological disorders, cancer, chromosome translocations, and degenerative neuromuscular conditions. Three themes were generated pertaining to parental experiences: the need for a streamlined pathway, listen to us, believe us, partner with us, and the power of an accurate diagnosis.

3.2 | Theme 1: The need for a streamlined pathway

Parents consistently report the challenges of navigating a pathway to genomic testing and services, with few experiencing a direct route. Parents found that both general practitioners (GPs) and pediatricians were unsure of how to refer their child appropriately for genomic testing or to genetic services. Positive referral experiences were identified when children were managed by a single, informed HCP. Parents identified that the assistance of a designated healthcare worker might assist fast-tracking access to genomic testing, co-ordinate and support them at medical appointments, support families during wait periods and assist them to engage efficiently with support services.

Diagnostic delays were attributed to numerous factors, including misdiagnosis, lengthy specialist wait times, extensive invasive investigations, and unclear referral pathways. Families living in rural or remote areas felt these delays were exacerbated. One parent suggested implementing an investigation “cutoff” before moving directly to genomic testing to minimize wait times, reduce child trauma, and alleviate financial burdens on families. Participants expressed frustration that genomic testing was not used initially, and,

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in some instances, genomic testing was needed for accurate diagnosis. Parents describe having to fight for access to genomic testing and services.

See Table 1 for illustrative quotes relevant to these subthemes.

3.3 | Theme 2: Listen to us, believe us, partner with us

The absence of designated healthcare workers to support families often left parents managing their child's care. This role included care coordinating and initiating or managing communication with HCPs. Parents describe having to "fight" for appointments with specialists, follow-up appointments, and testing and emphasized the need to advocate for their children, increasing their "case load" further. Some parents felt they were responsible for gathering information about their child's condition and sharing this with HCPs. Others noted the benefits of continuity of care in reducing the mental and emotional load they faced from acting as a case manager. A consistent team meant they did not have to continue retelling their child's story. Acting as managers heightened psychological stress for parents already coping with caring for a chronically ill child. Compounding this psychological stress parents often felt patronized, dismissed and like they were making things up through interactions with HCP's.

Furthermore, some parents also experienced social isolation because friends and other members of the public, on top of the health

TABLE 1 Illustrative quotes for subthemes demonstrating theme 1; Parental needs for a streamlined pathway to genetic services.

Theme 1 – A streamlined pathway		
Diagnostic delay	Misdiagnosis	"[Misdiagnosis] was a life-threatening condition, so I'm sitting here thinking [child] had this life-threatening condition…Three months later it came back, so 2 1/2 years down the wrong path. Back to the drawing board, another wait to see a geneticist." Participant 18 (Mother)
	Wait times	"The longest wait we had was the genetic testing itself, waiting for an appointment and waiting for the result of that." Participant 14 (Mother)
	Standard tests/cut offs	"From the start I think once you've executed the standardized testing through [Health service] there should be a cut off. No child or no patient should have to endure CT scans, blood tests, bone scans, lumbar punctures, you know, biopsies or MRIs multiple times. You know, and they should have a limit… an automatic jump to genetics…to minimize the effects the hospital treatment has and minimize the pain on the families, like financial pain." Participant 6 (Father)
HCP awareness	Referrals – who/what/when/where	"I thought she would be all across it, but she clearly didn't have a clue in terms of that side of things so that would be good, to have the pediatricians trained in knowing more about how to navigate the system and referring us earlier rather than waiting for so long." Participant 5 (Mother)
	Rare conditions	"Probably more doctors willing to look outside the box, … you know. It's the case of well, I am going to look at this, but not that, and not see the whole picture to do with the child." Participant 10 (Mother)
Stewardship	Blind navigation	"So [name of diagnosis] is what we left the appointment with. There was very little help. We looked at that and sort of threw our hands in the air and went well, what do you do with that?" Participant 6 (Father)
	Co-ordinating care	"Looking at other parents, like from outer areas, they don't have a very good time at all. We've pushed them to link with the social worker at the hospital so they can coordinate all the appointments, so they only have to come in for 1 day or 2 days for all the appointments for their specialist. Some are having a good experience, some aren't." Participant 8 (Mother)
	Support after diagnosis, during wait times and between visits.	"…if someone gets a diagnosis this severe, maybe a support worker, maybe a [Support service] person, someone, immediately, the same week if not the same day…someone comes to your house and talks you through it. Even if they don't know how to talk about the medical terms. If they are able to, procedure wise, able to direct the parents, it would make a lot of difference." Participant 11 (Mother)
	Accessing support services	"Accessing [Support service] is a nightmare. [Support service] don't recognize any genetic condition unless it's well publicized, so our son actually doesn't fall under the [Support service] for his syndrome or anything…they actually changed the label of his diagnosis…What they did is they changed it to a more overarching syndrome like muscular dystrophy…which totally and utterly did not describe our son at all. So, in them doing that we've actually lost funding…" Participant 6 (Father)

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system, did not understand what they were going through and could not provide support.

Crisis periods were also described, when parents were seeing multiple specialists and having numerous investigations around the same time. This was a confusing and difficult period for participants to recollect. These crisis periods and extra roles and responsibilities led to many parents feeling emotionally fatigued. Because of these challenges, psychological support provided by the health system was suggested by several parents, to assist them throughout their diagnostic journey and beyond.

Some parents reported how powerful and empowering it was when HCP's joined families in advocacy. These HCPs were instrumental in campaigning for access to genomic testing. Parents who were listened to were able to engage in partnerships with HCPs that fostered two-way learning about the condition and how to best support families grappling with rare conditions. Table 2 summarizes illustrative quotes for these subthemes.

3.4 | Theme 3: The power of accurate diagnosis

Post-diagnosis parents described positive experiences including validation, closure, and information to seek the supports they needed. Having the validation of a diagnosis legitimized parental concerns, meaning that they were taken more seriously by HCPs. Additionally, the closure afforded by a diagnosis reduced the guilt and self-blame parents had experienced.

Even though some children received difficult diagnoses, parents acknowledged that it allowed for acceptance and a plan for moving forward. They had a direction that allowed them to move on with their lives emotionally and practically. A diagnosis also helped families by enabling access to coordinated care, access appropriate support services, and connect with other families through online social support networks, including Facebook and WhatsApp groups. These connections facilitated valuable information sharing. Parents found support networks offered more current advice than medical professionals. Some participants experienced the greatest support from these groups, highlighting their importance in improving care for families with rare or undiagnosed conditions. Illustrative quotes for these subthemes are summarized in Table 3.

4 | DISCUSSION

The findings of this study reiterate aspects of the rare condition family experience previously reported including diagnostic delays, lack of awareness, coordination gaps in care, significant emotional and psychological impact on parents, insufficient government support, and the validation from an eventual diagnosis (Anderson et al., 2013; Currie & Szabo, 2019; Graungaard & Skov, 2007). However, we consider parents' experience in the context of growing accessibility of genomic testing and highlight factors that may reduce diagnostic delays and streamline access to genomic testing and services. These factors include promotion of genomic testing as a frontline test for suspected rare condition patients including educating HCPs, particularly GPs and pediatricians, and the development of models of care for rare condition families, supported by genetic counselors.

Parents valued a streamlined pathway to genomic testing to accelerate a diagnosis and alleviate additional emotional trauma and misdirection experienced throughout the diagnostic odyssey. Next-generation sequencing has improved diagnostic yields for rare disorders (McDonagh et al., 2021; Stark & Ellard, 2022; Turro et al., 2020). Furthermore, access to genomic testing has favorable effects on parents' emotional well-being, financial stability, family dynamics, decision making, and social connections (Wu et al., 2020). Compared with conventional care pathways, health economic analyses have found genomic sequencing less expensive and more cost effective (Stark et al., 2017, 2019; Wu et al., 2022). In multiple clinical settings, diverse stakeholders report the importance of a definitive genomic diagnosis for overall family wellbeing, the implications for clinical management, and the ability for HCP's and parents to register for notifications of recent advances in the treatment and management of the condition (Best et al., 2020; Goranitis et al., 2020; Pollard et al., 2021). However, the successful implementation of genomics programs throughout health systems requires a range of factors including, but not limited to, access to genetic counseling and building HCP clinical expertise in genomic medicine (Alarcón Garavito et al., 2023; Stark et al., 2023).

GPs and pediatricians are recognized for their pivotal role in appropriately referring patients, coordinating care, providing psychological support, and advocating for rare condition families (Dudding-Byth, 2015; Elliott & Zurynski, 2015). Similarly, in this study, parents felt emotionally supported when they formed a partnership with their HCP, and particularly grateful when this catalyzed access to specialist services including genetics. Unfortunately, these experiences were not universal. While HCPs report feeling their main role in rare conditions is referring families (Carroll et al., 2016), parents expressed a desire for coordinated care from one single HCP, more knowledge and support, necessitating targeted genomics education tailored to rare conditions.

The need for broader education on rare conditions among HCPs is underscored by strategic action plans worldwide, exemplified by Pillar 1, "Awareness and Education" of the Australian National Strategic Action Plan for Rare Diseases and Priority 2, "Raising awareness of rare diseases among healthcare professionals" of the UK Rare Disease Framework (Bhattacharya et al., 2021; UK Rare Disease Collaborative, 2021). GPs report inadequate preparedness to care for patients with rare conditions and encounters with people living with rare conditions may be underrecognized by HCPs (Elliott & Zurynski, 2015; Vandeborne et al., 2019; Zurynski, Gonzalez, et al., 2017). Pediatricians have expressed a desire for resources such as specialist referral lists and online education modules to support their understanding of rare conditions (Zurynski, Gonzalez, et al., 2017). Despite the development of educational resources in recent years; for example, the National Organization for Rare Disease Continuing Medical Education platform and the Australian RARE Portal, awareness of resources remains

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TABLE 2 Illustrative quotes for subthemes demonstrating theme 2; Parents need HCP's who listen to them, believe them and partner with them.

Theme 2 – Listen to us, believe us, partner with us		
Parents as case managers	Lack of consistency in healthcare team	"When you get admitted to emergency, you see 10 doctors, you tell them your life story over and over again…You don't want to be the expert, you want to be taken care of by someone." Participant 7 (Mother)
	Advocacy: 'the fight'	"… [initial testing] hasn't shown up anything…which was a bit… disheartening… so I ended up fighting for another appointment to then get additional sequencing…Push to get tested straight away… just don't give up. You know maybe it's not this, but, yes, just push for that second lot of testing, that wider array of (genetic) testing…" Participant 5 (Mother)
	Information gatherers	"We were told to look up [Condition Name] on PubMed and search on the web to see if we could find anything on [Condition Name]. There was nothing printed out and given to us. It was just advice on where to look." Participant 6 (Father)
Parental psychological struggles	Constant stress of a sick child	"Just being anxious not knowing when he was going to get sick again, trying to juggle hospital, work and the other children. The uncertainty of never knowing when the next hospital visit was going to be." Participant 14 (Mother)
	Am I crazy? Patronized and dismissed	"… the tone was 'do you actually think there is something wrong, you poor thing, he's fine don't worry about it'… it was hard work to advocate and feeling crazy and trying and get more answers." Participant 14 (Mother)
	Crisis points	"I guess, initially, when it all started, this was everything to me – trying to find an answer. As time goes on you get exhausted." Participant 19 (Mother)
	Lack of support for parents' psychological health	"…the absolute lack of support to families is, I think, a real gap in the system. Basically, parents have to bottom-out and then end up at the GP asking for a mental health plan, and I think the system should be much more proactive in helping prevent, the family fall-out that occurs so often in this diagnosis, by automatically ensuring parents receive psychological support." (Participant 4, Mother)
HCP partnerships	The support of empathy and belief	"Probably listening sympathetic ears…When you go through the whole sort of system, the doctors really do believe that they know more than you, and that is just not the case. Out of the seven specialists that we see…five of them listen, actually listen to me. The first thing they say is, 'Hi what's going on tell me what you need?'… Maybe the doctors are more aware now of the parents are the go-to for information." Participant 9, (Mother)
	Advocate	"The physio and the health nurse, they were great in that they came to the pediatrician appointment with me to help relay their concerns as well, so it wasn't just me." Participant 5 (Mother)
	Mutual respect	"…more trust in the parent, yes. Put a bit more trust and equalization, isn't a word, but sympathy, not sympathy because we don't need pity… Partnership that's the word…Yes, how can we help this together." Participant 8 (Mother)

limited (Tumiene et al., 2022; Zurynski, Gonzalez, et al., 2017). Overall, this study suggests that upskilling HCPs should include appropriate test ordering and interpretation, referral to genetic services, psychological and emotional support needs of rare condition families, early intervention recommendations, and connecting families with rare condition support networks. Approaches to boost HCP confidence and skill in genomic testing emphasize the importance of establishing relationships between genetics HCPs and primary HCPs through new service delivery models, rather than educational resources alone (Carroll et al., 2021; Crellin et al., 2019; Shelton & Whitcomb, 2015). Thus, genetic counselor supported models of care may facilitate genomic test ordering, and relationships between genetic services and HCPs that facilitate education.

Parents of children diagnosed with rare conditions serve as case coordinators (Baumbusch et al., 2019), and this was echoed in our study for parents prior to and following a genomic diagnosis. The strain of this role was further exacerbated by other challenges including identifying and accessing external services and lack of emotional and psychological supports (Baumbusch et al., 2019; Dellve et al., 2006; Khair & Pelentsov, 2019; Pelentsov et al., 2016; Picci et al., 2015). Genetic counselors are ideally placed to support rare condition families with multiple areas of expertise; discussing consent, testing options, providing information on recurrence risk and reproductive options, providing emotional support and facilitating psychological adaptation (Boycott et al., 2017), supporting family dynamics (McGlynn & Langfelder-Schwind, 2020), and promoting referral to appropriate supports. Furthermore, genetic counselors are trained in empowerment, shared decision making, and often play a vital role in building public awareness and trust (Veach et al., 2007). These attributes are recognized as essential in the promotion of equitable care pathways for rare condition families (Adachi et al., 2023) and uniquely position genetic counselors to create partnerships necessary for ongoing coordinated care.

Embedded genetic counselor models of care, initially reported in oncology services, have demonstrated increased referrals to

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TABLE 3 Illustrative quotes for subthemes demonstrating theme 3; The Power of an Accurate Diagnosis.

Theme 3: The power of an accurate diagnosis	
Validation	"It just confirmed, you know, as a relief and knowledge that there is something going on. I mean, I knew there was something going on, but having a name for it and being able to say, okay, this is a rare genetic condition, that's what you had to explain to other people, it was a massive release." Participant 5, (Mother)
Closure – Acceptance and moving forward	"I guess a downside of getting the diagnosis is you have to go through that grief, but if that's done in a supportive way, it helps you to move through that and progress." Participant 4 (Mother) "It felt like we were more acknowledged that yes, there is something, and we can put a name to it, and from there we were able to accept it, move forward, and get therapies and help him in the future." Participant 14 (Mother)
Information to seek out appropriate support groups, and connect with other families	"Did the diagnosis change anything? Absolutely not. It's just a label. It has done nothing to actually help him. What actually has helped us a lot was the ability to have the name, or the label then be able to link with other people." Participant 6 (Father)
Opened access to services	"In terms of funding, to be able to say, look this is a diagnosis, and we can access funding without too many questions." Participant 5 (Mother)

genetic services, reduced wait times, and improved access to testing, thus expediting treatment decisions (Bednar et al., 2017, 2019; Kentwell et al., 2017; Pederson et al., 2018; Rana et al., 2021; Senter et al., 2017). Recent implementation of similar models in pediatric settings has shown enhanced access to testing and personalized management, with high acceptability among patients and clinicians (Yanes, 2023). Of relevance, a recently published protocol proposed a model of care for embedding genetic counselors into primary care settings (Slomp et al., 2022). Addressing wider barriers to embedded models of care including, workforce planning, managerial support, and ongoing clinical genetics supervision may support future implementation (Quinn & Mazur, 2022). Our study highlights major health system gaps which inhibit access to genomic testing as well as adequate support services. We suggest that genetic counselors may be well suited to address these gaps.

4.1 | Practice implications

This study highlights that the challenges and needs of families with rare conditions align closely with the inherent skills of genetic counselors. Studies like this provide evidence for greater integration of genetic counselors into non-genetics general and specialist clinics. These roles could involve creating and delivering educational resources for HCP's and fostering relationships with them to facilitate direct access to genomic testing, care coordination, and long-term localized support. Genetic counselor led research may further establish the efficacy, feasibility, and cost-effectiveness of embedded models of care for the health system. Additionally, research in multiple settings will need to consistently demonstrate that patient reported outcomes are comparable in embedded genetic counselor models and clinical genetics services to promote wider implementation.

4.2 | Limitations

The participants in this study represent specific geographical locations (two states with different health services models). Despite these differences, the experiences reported by families were similar. Because there was a limited number of rural participants, there may be further challenges for these participants that were not captured in this study. Additionally, most of the participants were mothers, which may be reflective of their role as the primary carer, however, this limits the paternal perspective. Families who experienced more protracted diagnostic odysseys may have been more likely to participate.

4.3 | Suggestions for future research

As genomic testing becomes more accessible and economically feasible, evaluation of non-genetics HCP views about system barriers to the implementation of frontline genomic testing and review of referral education practices needs to be considered as part of national action plans for rare conditions. Our study highlights the value of further exploring genetic counselors embedded models of care for general and pediatric health services.

5 | CONCLUSION

A genomic diagnosis empowers families to make informed decisions about their child's health and identify relevant services and supports. Parents of children with rare conditions experience prolonged emotional and financial stress during their journey to genomic diagnosis. Therefore, access to genomic testing as a frontline option is crucial. Widespread testing will necessitate ongoing education of primary

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HCPs and warrants consideration of genetic counselor embedded models of care. Cumulatively, such interventions have the potential to facilitate the early identification of patients with rare conditions, improve access to an accurate diagnosis, and provide essential coordinated, medical, and psychological care for vulnerable families.

AUTHOR CONTRIBUTIONS

Aideen McInerney-Leo devised the project, the main conceptual ideas and outline. Courtney K. Wallingford, Molly Krause, and Aideen McInerney-Leo designed the investigation, wrote the protocol and ethics submission. Aideen McInerney-Leo, Heather Renton, Tatiane Yanes, Chris Jacobs, and Gemma Brett critically reviewed the protocol and interview guide. Courtney K. Wallingford and Molly Krause conducted interviews. Courtney K. Wallingford, Molly Krause, Aideen McInerney-Leo, and Amy Clark analyzed the data and interpreted results. Amy Clark, Courtney K. Wallingford, Molly Krause, and Aideen McInerney-Leo wrote the manuscript. Heather Renton, Tatiane Yanes, Chris Jacobs, and Gemma Brett critically revised the manuscript. Authors Courtney K. Wallingford, Molly Krause, Amy Clark, and Aideen McInerney-Leo confirm that they have had full access to all the data in the study and take responsibility for the integrity of the data and accuracy of the data analysis. All the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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CONFLICT OF INTEREST STATEMENT

Courtney Wallingford, Molly Krause, Amy, Clark, Heather Renton, Tatiane Yanes, Chris Jacobs, Gemma Brett, and Aideen McInerney-Leo declare they have no conflict of interest.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author and subject to ethical approval from the ethics committee that approved the original study. The data are not publicly available due to privacy or ethical restrictions.

ETHICS STATEMENT

Human studies and informed consent: Ethics approval was obtained concurrently in Victoria and Queensland through the Royal Children's Hospital in Melbourne, Victoria (HREC Approval: HREC/58138/RCHM-2019 RCH reference: HREC 2019.250) and through the University of Queensland Human Research Ethics Committee (HREC) (UQ/AMO3561). The HREC approval for Queensland was further ratified and approved by the University of Technology Sydney HREC (UTS/ETH19-C0004).

Animal studies: No non-human animal studies were conducted by the authors of this article.

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