

ORIGINAL ARTICLE

The experiences of families receiving a diagnosis of 22q11.2 deletion syndrome in Ireland

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Email: emma.odonoghue7@gmail.com**Abstract**

Families in Ireland often wait over 1 year to see a genetic counselor (GC). This qualitative study aimed to explore the views of families who received a diagnosis of 22q11DS in Ireland regarding the need for timely access to genetic counseling at the point of diagnosis. Twenty participants were recruited through the '22q Ireland' support group, giving a response rate of approximately 10% of the total support group members. Semi-structured interviews were conducted online and by telephone which explored experiences of receiving diagnoses, medical care, genetic counseling, mental health, and coping with the diagnosis. Interviews were transcribed verbatim and analyzed using thematic analysis. The experiences of 20 participants were classified into five main themes: Receiving Diagnosis, Interactions with Healthcare Professionals (HCPs, excluding GCs), Medical Care, Information, and Impact of Condition. Participants reported receiving diagnoses for their children in a sub-optimal manner due to inappropriate settings and insufficient information, support, and pre-test counseling. Parents reported feeling responsible for managing their child's complex and fragmented medical care. Participants reported insufficient empathy and little awareness of 22q11DS among HCPs. Participants perceived genetic counseling to be associated with family planning and reported delayed, if any, access to services. Mental health was a particular worry among participants. Conferences about 22q11DS are the main source of information for parents. Participants reported a range of emotions after diagnosis and described the family impact. The findings suggest both an association between HCPs' poor understanding of 22q11DS and the perceived lack of empathy from HCPs and fragmented medical care. There is an identified need for advocacy of the GC profession in Ireland to support these families. Increased awareness of 22q11DS among HCPs and the development of a coordinated care pathway for 22q11DS, with timely access to genetic counseling, may improve care and lead to better outcomes.

KEYWORDS

communication, complex disease, genetic counseling, genetic testing, lived experience, mental health, multidisciplinary

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1 | INTRODUCTION

22q11DS is the most common microdeletion syndrome (Hercher & Bruenner, 2008) with a frequency of approximately 1 in 4000 live births (Corvin, 2011; Du Montcel et al., 1996; Lawlor et al., 2017). 22q11DS, which includes Di George syndrome and Velocardiofacial Syndrome (VCFS), is a complex, multi-system disorder with over 180 age-dependent clinical features (Habel et al., 2014; Martin et al., 2012; Shprintzen, 2008). In infancy, acute medical problems including congenital heart defects, cleft palate and feeding difficulties, immune disorders, and hypocalcemia are observed (Campbell et al., 2018). In school years, cognitive, behavioral, and learning difficulties are common, with extra learning supports often required. In adolescent and adult years, there is potential for development of psychiatric disorders, including Attention Deficit Hyperactivity Disorder (ADHD), anxiety, depression, and schizophrenia (Campbell et al., 2018; Habel et al., 2014), with 52% of patients in a longitudinal study of patients with 22q11.2DS having a diagnosis of ADHD (Campbell et al., 2018). While not every person with 22q11DS will develop psychiatric illness, there is an increased risk of 25 times the general population of developing schizophrenia (Corvin, 2011; Shprintzen, 2008). One in every 4–5 individuals with 22q11DS will develop schizophrenia (Martin et al., 2012).

Following a diagnosis of 22q11DS, families in Ireland often wait over a year to see a genetic counselor (GC) and over 2 years to see a consultant geneticist (National Waiting List Data, National Treatment Purchase Fund, 2021). The recent introduction of a specialized multidisciplinary pediatric clinic for 22q11DS has helped to alleviate some issues for parents. However, there is currently no integrated involvement of GCs in conjunction with this clinical service.

It is recommended that families and individuals with a diagnosis of 22q11DS are offered genetic counseling (Driscoll et al., 1993; Greenhalgh et al., 2003; Liu et al., 2000; Ward et al., 2022). A study examining the experiences of families receiving a diagnosis of 22q11DS with a focus on genetic counseling has not previously been carried out in the Irish context. Ireland has a lower number of GCs per head of population in comparison to European countries of similar population (Abacan et al., 2019), which may contribute to families having different experiences of access to services of a GC when compared to studies carried out in other countries such as the UK, USA, and Australia. Genetics services are not well-understood within the Irish healthcare system and there is currently no cohesive genomics strategy in place, although a National Genomics Strategy is under development in 2022. One Irish study investigated parents' experiences of communication at diagnosis around the psychiatric issues associated with 22q11DS, and the potential benefits of a 'psycho-educational' program for families (Alugo et al., 2017), which gives a good background to the current situation in Ireland. However, genetic counseling and the benefits that timely access to GCs may have for families with 22q11DS in Ireland were not examined in the study. This study addresses a gap in the current literature and investigates the potential benefits of timely access to GCs for families with 22q11DS in Ireland.

What is known about this topic

There are potential benefits of a psycho-educational program for families in Ireland who receive a diagnosis of 22q11DS for their child (Alugo et al., 2017).

What this paper adds to the topic

This study is the first to explore the experiences of families receiving diagnoses of 22q11DS with a focus on genetic counseling in Ireland, a country with fewer GCs per capita than other European countries of similar size population (Abacan et al., 2019). The study identifies a need for the advocacy of the GC profession in Ireland in the form of adequate staffing levels and resourcing of services to support families receiving a diagnosis of 22q11DS.

Individuals within the rare disease community experience many years of misdiagnoses or awaiting diagnoses, a phenomenon termed the 'diagnostic odyssey' (Vandeborne et al., 2019). Multiple appointments and referrals to different specialisms are reported by families with 22q11DS, such as cardiology, genetics, immunology, pediatrics, and speech and language therapy in particular (Campbell et al., 2018). This may cause emotional and financial burden to families (Pelentsov et al., 2016; Reilly et al., 2015; Vandeborne et al., 2019).

Parents of children with 22q11DS from Ireland have reported feelings of frustration due to a lack of support and information received from HCPs (Alugo et al., 2017), which echoes recurring themes reported in the wider rare disease community. Parents' dissatisfaction with HCPs' levels of knowledge and support may direct them to find other sources of information (Alugo et al., 2017; Cuthbert et al., 2019; Vandeborne et al., 2019), which are more meaningful to them, such as support groups and online forums (Rizzo et al., 2020). This suggests that HCPs are not providing enough psychological support to families receiving a diagnosis of 22q11DS and suggests that more input from GCs at the stage of diagnosis would be welcomed (Rizzo et al., 2020).

Furthermore, the 22q11DS phenotype is highly variable from person to person (Campbell et al., 2018), even between members of the same family. This variability, along with the age-dependent clinical features of 22q11DS, may heighten the impact of HCPs lack of awareness of 22q11DS due to the complexity of the condition. While non-genetics HCPs can help manage the symptoms associated with 22q11DS, parents coping with a diagnosis of 22q11DS in their child have particular needs that non-genetics HCPs may not have the resources or training to address (Adlington et al., 2019). Outpatient appointments are often too short to explore the worries of the parent and rather focus on the physical symptoms of the child, leaving the parent to adapt and cope on their own or with the help of support groups.

As GC training is rooted in Rogerian-based counseling theory and other theories such as Family Systems theory (Bowen, 1978), GCs are likely to be well-placed to provide psychosocial support to families receiving diagnoses of 22q11DS (Austin et al., 2014; Rizzo et al., 2020) and other rare conditions. Acceptance and coping with uncertainty can be exceptionally difficult for parents dealing with a new diagnosis of 22q11DS in the family (Broley, 2013). The use of Acceptance and Commitment Therapy (ACT), a form of a cognitive and behavioral therapy (CBT) focusing on mindfulness and acceptance through the use of metaphor and value-focused exercises, has been outlined in a genetic counseling case study with a parent of a child diagnosed with 22q11DS who is experiencing intrusive worrying thoughts (Broley, 2013). While the benefits of this technique have not been studied in good-sized samples, this short-term intervention is an example of the family-oriented training and counseling skills that GCs may possess which may be well-suited to supporting parents and families receiving a diagnosis of 22q11DS.

As clinical genetics departments cannot facilitate appointments for all individuals undergoing genetic testing in other specialties and as mainstreaming is introduced, the communication of 22q11DS diagnoses to families in the UK is increasingly being done by non-genetics HCPs such as pediatricians (Adlington et al., 2019). This is similarly occurring in the rare disease setting in general, where another study reports that pediatricians are more likely to communicate genetic diagnoses to families in the UK than compared to the US, where this is predominantly carried out by genetics specialists (Cuthbert et al., 2019). This study reports a higher proportion of UK participants (39.2%) experiencing dissatisfaction with the disclosure of diagnoses and information for psychiatric risks in children with rare disorders, as compared to US participants (26.4%; Cuthbert et al., 2019).

A feature which is specific to 22q11DS is the increased risk of the onset of psychiatric symptoms. Parents report dissatisfaction with the information provided by HCPs around the psychiatric risks associated with 22q11DS (Cuthbert et al., 2019; Hercher & Bruenner, 2008; Martin et al., 2012). Satisfaction was found to be predicted by several factors including face-to-face communication, communication by genetics specialists and receiving support (Cuthbert et al., 2019). It has been recommended that GCs communicate the risks of psychiatric conditions to families (Reilly et al., 2015) and has been reported by GCs themselves that it is preferable to disclose information regarding the psychiatric risks to parents (Martin et al., 2012). Not disclosing the mental health (MH) risks associated with 22q11DS by HCPs is likely to be unhelpful as multiple studies have reported that parents find this information from online sources, rather than from HCPs (Cuthbert et al., 2019; Hercher & Bruenner, 2008; Rizzo et al., 2020), which could potentially be upsetting for parents.

The disclosure of 22q11DS diagnoses raises many complex issues. The literature to date suggests that communication of diagnoses may best occur via genetics specialists such as GCs, in a sensitive and timely manner accompanied by the offer of support or appropriate signposting, in order to provide the best outcomes for

the individual and family. Influencing factors in parents' experiences of disclosure of diagnoses in the literature include HCPs' low awareness of 22q11DS and the educational needs of HCPs to provide improved information and support, including the associated risk of psychiatric illness. For 22q11DS specifically, perhaps the complexity of the condition (Rizzo et al., 2020) may further contribute to HCPs' low awareness (Vandeborne et al., 2019) and parents' perceptions of lack of support and information from HCPs (Cuthbert et al., 2019; Pelentsov et al., 2016). The literature demonstrates that the provision of information and support to families at the time of diagnosis needs to be improved, both in Ireland and other countries. There is a lack of sufficient data in the Irish context, particularly around how the diagnosis of 22q11DS and associated psychiatric risks are communicated to families, how families cope with and adjust to such news, and how this could be improved. This study was designed to fill these gaps and explore the needs of these families, in relation to the research question 'what are the views of families who received a diagnosis of 22q11DS in Ireland on the need for timely access to GCs at the point of diagnosis?'

2 | METHODS

2.1 | Study design

A cross-sectional semi-structured interview-based qualitative approach was chosen to retrospectively explore participants' experiences of receiving their child's 22q11DS diagnosis. Ethics approval was sought from the Cardiff University School of Medicine Research Ethics Committee in July 2020 and granted in August 2020 (reference SMREC 20/76).

2.2 | Participants

The target population was parents of children diagnosed with 22q11DS. The inclusion criteria for participation were; (a) parent of a child diagnosed with 22q11DS, (b) fluency in English, (c) over 18 years of age, and (d) able to consent to participate in the study. The exclusion criteria were; (a) not a parent of a child diagnosed with 22q11DS, (b) not able to converse in English, (c) under 18 years of age and (d) not able to consent to participate in the study. The target sample size was 12–15 participants to aim for data saturation for thematic analysis (Hill et al., 2005; MacFarlane et al., 2014). A purposive sampling strategy was employed.

Recruitment for the study was carried out through the '22q Ireland' support group. The researchers did not have a relationship established with participants prior to study commencement, with the exception of the 22q Ireland support group representative. The study was advertised by 22q Ireland to approximately 195 of its members via email and private Facebook group. Support group members who expressed interest in the study were provided with the Participant Information Sheet (PIS; see Appendix S1) and

consent form. The project advertisement was ceased when 20 participant interviews and data analyses were complete due to data saturation being achieved. Reasons for why other 22q Ireland support group members did not seek to participate in the study were not investigated.

2.3 | Procedures

Semi-structured interviews were conducted by EOD with participants from September to November 2020 by telephone and Cardiff University 'Blackboard Collaborate' video-conferencing. Interviews could not take place in person due to the COVID-19 pandemic, as regions of Ireland were under 'level five' lockdown during this period. While interviews conducted by telephone and Blackboard Collaborate did not capture non-verbal cues such as body language, these platforms had the benefit of allowing participants to participate in the study from the safety and comfort of their own homes and at flexible, convenient times, which was a particular advantage during the COVID-19 pandemic. Interviews lasted between 33 and 93 minutes (mean = 48). Repeat interviews were not conducted.

An interview guide was used to collect data during the semi-structured interviews (see Appendix S2). The interview guide was designed corresponding to topics identified during the literature review (Alugo et al., 2017; Cuthbert et al., 2019; Rizzo et al., 2020) and was also driven by the research question. The interview guide was not pilot tested prior to interviews taking place. Audio data were recorded and transcribed verbatim by EOD. Identifying details such as names and locations were omitted during transcription to maintain confidentiality. Field notes were also recorded by EOD. Transcripts were not returned to participants for corrections or feedback.

2.4 | Data analysis

Thematic analysis was carried out by EOD by following the six steps outlined by Braun and Clarke (2006). The coding of two sets of interview data were blindly second-checked by RR in order to ensure accuracy, to increase confidence in the analysis, and reduce researcher bias (Hill et al., 2005; MacFarlane et al., 2014). Any ambiguities in the coding data were addressed through critical dialog between EOD and RR. The analysis was informed by the roles of the researchers as GC student (EOD) and as GC with previous research undertaken with families with 22q11DS (RR), while practicing reflexivity to reduce any potential professionalization bias (Hiller & Vears, 2016). Codes were reviewed and formed into overarching themes as they became apparent. Themes were not identified in advance but were derived from the interview data. An Excel spreadsheet was created with a tab for each theme and each participant's codes listed across each column within each tab.

This was reviewed to decipher how codes belonged within themes and how themes related to each other. Codes were collated into overarching themes and subthemes, and a final thematic map was produced (Figure 1).

3 | RESULTS

3.1 | Sample characteristics and demographic information

Members of the 22q Ireland support group responded to the project advertisement by contacting the researcher with completed consent forms. Project advertisement ceased after 20 participants were interviewed, giving a response rate of approximately 10% of the total membership of the 22q Ireland support group. Sixteen participants (80%) were female caregivers and 4 participants (20%) were male caregivers. One parent interviewed was diagnosed with 22q11DS after receiving their child's diagnosis, while the other 19 parents did not have a diagnosis of 22q11DS (i.e., occurred de novo in their child). The years in which the diagnoses were received ranged from 1997 to 2018, spanning a range of 21 years. Half (50%; $n = 10$) of diagnoses were received when the affected child was between the ages of 3 and 6 years. Three participants in this study received 22q11DS genetic diagnoses for their child prenatally, resulting from the detection of abnormalities identified at the anomaly scan at approximately 20 weeks of pregnancy. Genetic diagnoses were most commonly disclosed by pediatricians (30%; $n = 6$), followed by clinical geneticists (25%; $n = 5$), cardiologists (20%; $n = 4$), fetal medicine consultants (15%; $n = 3$) and neonatologists (10%; $n = 2$). Twenty-five percent ($n = 5$) of participants were never referred to a Clinical Genetics service, that is, at the time of interview these participants had never seen a consultant geneticist or GC. Of the 15 participants who had been referred to Clinical Genetics by the time of interview, 50% ($n = 10$) were seen by a consultant geneticist and 30% ($n = 6$) were seen by a GC. One participant was seen by both a consultant geneticist and GC. Participant characteristics are displayed in Table 1.

4 | RESULTS OF THEMATIC ANALYSIS

Thematic analysis of the interview data identified five major themes: Receiving Diagnosis, Interactions with HCPs, Medical Care, Information, and Impact of Condition. Themes derived from the interview data and not identified in advance of the study being conducted. Several subthemes were identified within the overarching themes. These themes are presented in Figure 1 as a thematic map, where the dark-colored circles represent the overarching themes and the lighter-colored circles represent the subthemes. The size of the circles represents the prevalence of the subject in the interview data.



FIGURE 1 Thematic map

4.1 | Receiving diagnosis

This theme was central to each of the themes and subthemes. The way in which diagnoses were delivered to parents had an impact on their subsequent adaptation to their child's diagnosis and experiences of care. A large proportion of participants described their experience of receiving the diagnosis for their child as primarily negative, with many participants describing their experience of receiving the diagnosis as 'cold'. Many participants described receiving their child's diagnosis in inappropriate settings, such as at their child's bedside in hospital or by phone call, sometimes while at work or while driving. Seven of 20 participants were alone when they received the diagnosis.

P8: "I was in work and I just got a phone call and I was told there and then and that approach, to me, was completely and utterly wrong. I didn't know whether to break down, like, I couldn't break down 'cause I was in work which is not a place where you want that to happen"

As described in the above quote, not being given prior notice of a phone call to communicate genetic test results appears to have had a negative effect on the participant.

One participant was diagnosed with 22q11DS as a result of parental genetic testing following their child's diagnosis. Both diagnoses were communicated to the family over the phone, which felt to the participant to be an example of the lack of family-centered care and planning around the disclosure of diagnoses to the family.

P7: "We were told over the phone, em, that I had it and has passed it, like... over the phone again, I was just going home from work, I'll never forget it ... I was a bit bitter about it, it wasn't a nice way to tell me"

There was also a reported lack of appropriate information provided at diagnoses. Participants wished for HCPs to listen to their concerns, rather than giving a list of possible symptoms or worst-case scenarios. Eleven of 20 parents described how their child's diagnosis was given to them as 'Di George syndrome' and expressed this as a negative element of the disclosure of diagnosis. One parent described how being given the name of the condition as 'Di George syndrome' prevented them from initially finding a 22q11DS support group after diagnosis.

P18: "[HCP] called it Di George syndrome ... which is horrendous if you Google it"

TABLE 1 Participant characteristics

Participant code	Gender	Age of child at diagnosis	Diagnosis disclosed by	Referred to clinical genetics	Seen by GC or consultant
P1	Female	15 months	Geneticist	Yes	Consultant
P2	Female	15 years	Geneticist	Yes	Both
P3	Male	1 month	Neonatologist	Yes	Consultant
P4	Female	1 month	Neonatologist	Yes	Consultant
P5	Female	3 years	Pediatrician	No	N/A
P6	Female	4 years	Pediatrician	Yes	Consultant
P7	Female	6 years	Pediatrician	Yes	GC
P8	Male	6 years	Pediatrician	Yes	GC
P9	Female	3 weeks	Cardiologist	No	N/A
P10	Female	4 years	Pediatrician	Yes	GC
P11	Male	4 years	Pediatrician	Yes	GC
P12	Female	5 years	Geneticist	Yes	Consultant
P13	Female	5 years	Cardiologist	Yes	GC
P14	Female	3 weeks	Cardiologist	No	N/A
P15	Female	Antenatal	Fetal Medicine	Yes	Consultant
P16	Male	6 years	Geneticist	Yes	Consultant
P17	Female	6 years	Geneticist	Yes	Consultant
P18	Female	Antenatal	Fetal Medicine	No	N/A
P19	Female	4 months	Cardiologist	No	N/A
P20	Female	Antenatal	Fetal Medicine	Yes	Consultant

Parents also described a lack of psychological support at diagnosis and made suggestions about how having an HCP in a supportive role present at the diagnosis could be helpful. Many participants felt unprepared to receive results, suggesting a lack of pre-test counseling.

P4: "There's a gap there as to why [child] was getting the genetic test, even though [HCP] came in and [HCP] explained it to me, but it all sounded very run of the mill, normal, this happens all the time, kind of stuff"

While the majority of parents experienced the disclosure of diagnosis in a negative manner, some parents expressed praise for how their diagnosis was delivered, although acknowledging that receiving a diagnosis for your child is not a positive experience in general. Positive elements of how diagnoses were delivered included being prepared to receive a diagnosis, receiving the diagnosis in person and not being alone when receiving the diagnosis.

P12: "we had got a letter saying ... you have to come to the Genetic clinic on such a date, do not bring your child in, so I think before we walked in the door we kind of knew... something had changed here, you know"

4.2 | Interactions with HCPs

Interactions with HCPs were a major theme throughout the interview data as parents of children with 22q11DS have frequent interactions with many HCPs. For example, two participants described how their child has been under the care of many different doctors in a 12-year period. Participants described having to repeatedly explain the condition to HCPs due to a low awareness of the complexity of the condition among HCPs. Participants reported feeling lost when HCPs are not knowledgeable about the condition and described having to educate HCPs about 22q11DS.

P16: "we've dealt with ... probably up close to fifty medical doctors within the last twelve years"

P13: "My own doctor had to Google it in front of me and said 'I don't know what that is'"

When participants met HCPs with an understanding of 22q11DS, they expressed feelings of relief, hope, and enthusiasm. Participants felt that the naming of the condition affects the awareness between HCPs, with HCPs having a higher awareness of 'Di George syndrome' but a low awareness of the condition when referred to as '22q11 deletion syndrome'. Four participants expressed feeling that HCPs have

a lack of knowledge of 22q11DS in comparison to other conditions such as Down syndrome. Most participants remarked that the awareness of 22q11DS is increasing and this was largely attributed to the advocacy work of a local 22q11DS support group and the 22q clinic. Many participants described a lower awareness of the condition in local services.

P13: "you shouldn't have to fight to figure out the pathway ... like for stuff that is not so rare, there is a pathway if you're diagnosed with Down syndrome that people know about and understand"

P14: "I think the awareness has definitely been, especially through [support group chairperson] and the 22q group, like, it's skyrocketed, what she's done"

P20: "I don't expect to get the same service in the local hospital"

Participants also described experiencing a lack of empathy from HCPs, including not feeling listened to and their child's symptoms being minimized by HCPs prior to the diagnosis. Several participants expressed a desire for HCPs to be patient-centered and to focus on the parent's main concerns about their child. Several participants also described how some HCPs perceived the condition negatively.

P11: "The one thing that always irks parents, I think, is a, a professional sees [child] for a snapshot, whereas we as parents are seeing him the whole time and, and sometimes with some consultants and different things, you do feel like ... you really have to push your side to, to get them to listen or hear you"

P20: "The [HCP] we had was very much, em, focused on maybe some of the more negative outcomes with mental health or problems like delinquency"

Some participants described feeling that HCPs insinuated they were being overly cautious.

P5: "One doctor wrote back to my GP and said 'the mother is, like, overly anxious' and you know, something to reference me being, like, nearly neurotic"

P13: "You know, 'first time mum', 'bit anxious', like"

4.2.1 | Genetic counseling

In this study, six participants had an appointment with a GC after their child's 22q11DS diagnosis. Most participants described not understanding what genetic counseling is. Some participants also reported HCPs having a low awareness of the genetic counseling

profession and services, a factor which may explain the low referral rate for participants in this study.

P5: "I actually asked one of the [HCP], and she said 'you can't get that done in Ireland, em, I think you have to go away for that, I've never, I've never heard of it ... it's not something that's really done here'"

There was an expectation among some participants of genetic counseling as psychological counseling, which participants felt was not what their experience of was attending a GC. There was also a strong perception among participants of the association of genetic counseling with family planning. One participant declined an appointment with a GC due to their understanding of genetic counseling as family planning, as they had completed their family.

P16: "I pigeon-holed the whole genetic counselling as being about ... whether you should have another baby and all that and surely it's ... a bit vastly wider than that, but just because we've never had it, I don't even have a clear view on what genetic counselling is"

P19: "I got a call saying that they had, that I was on the top of the list for genetic counselling and there was a cancellation and could I come in for a chat. At that point I'd had a hysterectomy so I was like 'is genetic counselling just about me planning another family? ... well, that's not going to happen so you can take me off your list'"

Participants who did attend a GC for genetic counseling had diverse experiences. Several participants described having high expectations for genetic counseling and a desire to know how their child's deletion would affect them, rather than receiving generic or scientific information.

P13: "The word counselling, you know, it isn't really and I certainly expected a lot more than what I got. I hoped, I thought I'd get an understanding more of the diagnosis ... it didn't give me any understanding of what we were dealing with or what we were potentially dealing with which is what I would have, thought I might have been"

Participants were aware of the long waiting lists to access clinical genetics services and the impact of this delay on parents, particularly where parents had already educated themselves on the condition.

P6: "We basically lost a year and a half for [child] because ... he had to wait for his genetic appointment ... for ten months and freaking out because you don't know what applies to your child what you read online and the more you read online the scarier it can get"

Participants also described how the timing of appointments with clinical genetics may have been more useful prior to the testing being done, or shortly after diagnosis.

P10: "Five months after the diagnosis we got an appointment ... by that stage we had done a lot of research ourselves"

Many participants commented how follow-up GC appointments should be offered similarly to other medical appointments, and particularly about the need for access to GCs at different life stages. Several participants mentioned the benefit that attending a GC may have for teenage children with 22q11DS. Parents described not feeling equipped to tell their child about their condition. Two participants also mentioned the particular relevance of psychiatric genetic counseling for 22q11DS.

P2: "I also think ... that kind of branch of, that new branch of genetic counselling, the psychiatric genetic counselling, is particularly relevant to 22q because of the mental health"

Overall, participants described how attending a GC can provide reassurance, an opportunity to ask questions and to gain an understanding of the condition. Several participants remarked how access to a GC appointment should be available to everyone receiving a genetic diagnosis.

P20: "... how important it was for us to receive that genetic counselling and that ... I just think everybody should be... an option, like, everybody should have that. It should be part of a diagnosis"

4.3 | Medical care

Many participants described their experiences of fragmented care, long waiting lists and barriers to timely access to medical services. This was largely attributed to a lack of resources within healthcare services and convoluted referral pathways. Several participants also noted the impact of the COVID-19 pandemic on the increasing length of waiting lists. The reliance on private healthcare was mentioned by many participants, further highlighting the lack of adequate resourcing of medical care.

P9: "We've had twenty-three years of waiting lists. Even ... when [child] had the [surgery] she was five years on the urgent list"

Many participants described their experience of healthcare departments working in isolation and expressed frustrations with HCPs not considering their child's different presentations as a whole. An example given by several participants was the use of growth charts

which are not specific to children with 22q11DS. Several participants also expressed frustration with the 'wait and see' approach taken by some HCPs.

P17: "It's all very compartmentalized, you know, cardiac only look at cardiac"

4.3.1 | Burden of responsibility on parents

Participants in the study expressed how the responsibility for their child's medical care is largely placed on the parents. Nearly all participants described having to fight for access to resources and advocating for their child's needs. Participants reported staggering numbers of appointments, particularly in the earlier years following diagnosis. Participants described the physical burdens that this has on parents. However, the number of appointments tended to decrease with age. Participants also described having to source information and become very knowledgeable themselves about the condition.

P1: "you're waiting or you're fighting for something, you know what I mean, you're, you're constantly chasing something"

P11: "we had to learn a lot about the medical world ... without having any background in it whatsoever, to overseeing [child]'s overall healthcare"

P14: "you need to be quite proactive, or if you sat back and waited for all your appointments to come in the post they wouldn't necessarily come ... I used to say I needed a PA for all of his appointments"

4.3.2 | 22q Clinic

Approximately 75% ($n = 15$) of participants attended a specialized 22q clinic which was set up in 2017, while the remainder of the participants' children had transitioned to adult care before the clinic was set up. The 22q clinic involves both first referral appointments, covering a full medical history from pregnancy to the present day, arranging blood tests and referral to other specialties as needed, and review appointments which are shorter and include a review of results and any new needs identified. Recently, teens also have the opportunity to meet a consultant involved with an adult 22q clinic to aid transition. Overall, there was strong praise for the 22q clinic. Participants expressed a feeling of relief and comfort of having access to HCPs who are familiar with the condition.

P11: "I remember coming out of that appointment and ... felt such a relief, such a weight off our

shoulders knowing there was somebody that was, has it all under control”

Participants described the necessity of having a specialized clinic for 22q11DS as not every hospital will have the same level of expertise with 22q11DS. Another significant benefit of the 22q clinic was the reduction in the number of appointments to be attended and helping to resolve fragmented healthcare, such as having annual bloods taken while at the 22q clinic which reduces parents having to attend multiple appointments in different locations for blood tests. One criticism was the length of time waiting to get an appointment after being referred to the 22q clinic. Some also commented on there being more of a focus on the medical side of 22q11DS than the psychiatric or behavioral side. One participant mentioned how the 22q clinic is in need of appropriate funding.

P8: “You're going everywhere, whereas, you know, having one natural base where you get seen by a number of professionals and you get seen in the one day is much handier, especially by professionals that understand the condition”

P10: “We're going to the 22q clinic, you know ... so we don't need to go see immunology every year ... Those sort of things are priceless, do you know, just the little bit of efficiency”

P13: “It's still pretty much volunteering and the graciousness of one doctor that's interested in it”

Participants reported how non-genetics HCPs in the 22q clinic are required to provide genetic counseling to parents due to the lack of timely access to GCs. One participant described how genetic counseling could be offered as part of the 22q clinic as it is something that every family could benefit from.

P14: “[genetic counselling]'s probably something that ... every child or every family can benefit from where, you know, some of the things could be kidneys or their eyes or whatever, you know, not every child has but, I suppose, that's something that everybody could benefit from, so it is certainly something that could be incorporated into the 22q clinic”

4.3.3 | Transition

The transition into adulthood and the uncertainty of the future was a concern for many participants. Some parents described their child being behind their peers. Many participants noted the overlap between pediatric and adult care needs for individuals with 22q11DS. Parents of adults with 22q11DS described how they attend multiple

adult hospitals for medical appointments and how this has led to a decreased quality in their care.

P12: “Now we're cardiac in [hospital A], we're ENT in [hospital B], we're immunology in [hospital C] as well, but now we're kind of ... we're in different hospitals, so it, so it's actually got worse again”

P16: “When [child] hits eighteen and whatever the transition is... I can see one of the, the really big things about 22q is that kids keep learning and keep maturing into their mid-twenties”

Empowering children with 22q11DS to speak on their own behalf in relation to their medical care was something that many parents were supportive of and eager to implement. The importance of children building a good relationship with their GP as they come into adulthood was also mentioned. Many participants also expressed that an adult 22q clinic is needed with the need for a focus on social and MH issues at this point. A need for access to a GC at this point in life was also mentioned. Many parents described concerns about their child with 22q11DS coming into childbearing age, which further highlights a need for better access to genetic counseling at different life stages.

P9: “I do think they should be seen [by a GC] maybe a few times ... the original diagnosis and maybe a couple of times after that and then maybe later on ... when they're transitioning into adulthood”

4.4 | Information

Participants in the study sought certainty about the condition and their child's future. Participants also reported that being given the diagnosis as ‘Di George syndrome’ directed them toward different streams of information. Nearly every participant described confusion caused due to the naming of the condition. One participant described how it took many years to find the support group due to the diagnosis being given as Di George syndrome.

P9: “The group would have been together a few years before we even found them and then again it came back to the name, you know, because we had been given the diagnosis of Di George”

P11: “if you Google Di George syndrome and you Google 22q it's very, very different... like, Di George when you're Googling it, it's very, very scary, like ... it's much older information... the prognosis isn't good”

4.4.1 | Mental health

This subtheme refers to the association of MH problems with 22q11DS, which was perceived as a particularly worrying element of the condition by most participants. Many participants could not recall the MH association being discussed at diagnosis. Most participants who received information about the MH association at diagnosis described how it was done in a negative or cold manner. Some participants were not informed of the MH association at all by HCPs and described being left to figure it out over the years. Many parents described reading and hearing material about the association of MH problems as overwhelming and distressing.

P1: "I remember when we were going to the first conference, and I remember saying to my husband ... we found it very upsetting to, I suppose [child] was only ... a baby or whatever, it was very upsetting to hear ... the bad stories"

P19: "The mental health thing was the thing that haunted me actually for the first couple of, em, I read so much about that"

Although the information was perceived by participants as overwhelming, all participants described the need for parents to be informed about the MH association. Participants reported a lack of understanding by HCPs of the link between 22q11DS and MH. Many participants expressed a desire to be equipped with knowledge in order to be proactive to best support their child.

P9: "The more aware a parent is, the better ... you can deal with the information, as they say, knowledge is power, like, but having to pick out the information, I think, can be ten times harder"

4.4.2 | 22q11DS support group

Participants valued the peer support they receive from a local 22q11DS support group. Most participants talked about the support group in terms of access to reliable information, as well as the emotional support it provides. The support group also provides information that parents can give to HCPs. Many participants described how conferences organized by the support group and the Max Appeal consensus document (an information resource developed by a UK-based 22q11DS support group) were the first sources of information about the MH association with the condition.

P14: "we have a support group, we have a private parents group, em, that we chat, so if we have issues, we can talk or... we all find that really, really useful, em, helpful to talk to each other, just to share ideas or to

see something, you know, are you the only one that's coming up with certain things"

P15: "that conference was the first time I really heard about [the mental health association] and got an understanding of it"

P8: "thanks to [support group reps], like, they kind of gave us a lot of information that we could then bring to doctors and ... the healthcare professionals to kind of explain a bit more about the condition"

4.5 | Impact of condition

The impact of the condition on their child's health was a significant source of worry and distress for participants.

4.5.1 | Coping with the diagnosis

Participants described the impact of receiving the diagnosis. Most participants described feeling overwhelmed, devastated, in shock and denial. Many participants described a desire to meet other families with the condition and expressed a comfort in knowing there are other families out there.

P4: "It was pure shock to be honest with you ... I kind of felt paralysed"

P18: "I was devastated, I really was ... I found it very tough going forward"

Some participants discussed differences in the acceptance of the diagnosis between female and male caregivers. Several participants expressed how fathers, in their experience, can find it harder to accept the diagnosis. Participants in the study who were mothers tended to attend conferences and participate with the support group more than fathers.

P11: "When I'd be talking to some of ... the fathers, you do feel that there is a tiny disconnect with how much is going on and I do think ... a fathers a little bit more likely to be a second-hand person, than, lots of the, the fathers I've met, some of them don't even want to mention the word 22q or don't want their kid to ever find out about it"

Feelings of guilt were often mentioned by participants during the interviews, particularly with mothers in relation to pregnancy. Several participants remarked on the lack of psychological support for parents and some expressed a desire for access to counseling. Nearly all

participants described a need for more support for families. Several participants expressed praise for parent workshops organized by support groups. Some participants described not wanting to focus too much on their worries about the future, particularly regarding the MH aspect and children growing into adulthood. In particular, these participants described 'stepping back' from information about MH when it became too overwhelming.

P19: "you can't allow yourself to go into that space of... worrying projectively down the road so I really try and... and not think about that, to be honest with you, at the minute"

4.5.2 | Impact on the family unit

The impact of the condition on the whole family was also discussed by many participants. Several participants reflected how having a sibling with 22q11DS makes siblings more understanding and less judgmental of others.

P1: "Siblings are just brought up to, to deal with living with the child and they're being asked from a very early age to be more patient, to be more understanding, to, you know, one rule doesn't apply to, one rule applies to the other"

However, one participant described how her child has decided not to pursue having children due to their experience of having a sibling with additional needs.

P18: "He would have huge resentment for ... having a sibling with a disability, you know ... he's just saying 'I will never have children'. I think he just, he has justified that there [will be] no children for him"

5 | DISCUSSION

This study identifies issues which are specific to families with 22q11DS in Ireland, but many of these have also been reported within the wider rare disease community. Parents bear a large responsibility for managing their child's complex care, receive little or no psychological support and experience a low level of understanding of their child's condition from HCPs. The understanding of and referral to GC services among non-genetics HCPs were perceived by participants to be low. A central finding was the negative manner in which the participants in the current study received their diagnoses. The main factors which contributed to these negative experiences were the inappropriate settings in which diagnoses took place and the lack of family-centered information and support. These findings support previous findings by Alugo et al. (2017). An Irish report released in 2007 by the National Federation of Voluntary

Bodies, entitled 'Informing Families of their Child's Disability', outlines best practice guidelines for delivering diagnoses to families (Harnett, 2007). This report outlined parents' poor experiences of receiving diagnoses for their child's disability. Recommendations made in the report include giving a realistic and hopeful outlook to parents and not focusing on the worst-case scenario. The report also recommends for the disclosure of diagnoses to be family-centered and to take place in an appropriate setting.

The findings of this study suggest that some participants may have had similar experiences to a wider set of parents of children with other disabilities as they were reported in 2007. This suggests that these best practice guidelines may not be implemented by all HCPs delivering 22q11DS diagnoses, which supports the more recent findings from Alugo et al. (2017).

A lack of pre-test genetic counseling was evident in many interviews due to participants' apparent surprise at receiving genetic test results. This apparent gap in understanding of the possible outcomes and limitations of testing suggests a lack of sufficient pre-test counseling, with some parents not being aware that a genetic test had been carried out and were not expecting to receive results. The importance of pre-test counseling for pediatric genetic testing has been recognized for many years (Palmer et al., 2012) by appropriately trained HCPs or with the assistance of GCs. Lack of time and significance dedicated to pre-test counseling by non-genetics HCPs may suggest a lack of understanding of the impact of a condition such as 22q11DS on families. However, it must be acknowledged that it cannot be possible for HCPs to prepare families for all possible diagnoses and uncertain results which may result from broad genetic testing such as microarray testing, and therefore preparing parents for a range of possible results and diagnoses is inherently difficult in pre-test counseling. The issue is also raised regarding the need for adequately resourced clinical services which can feasibly allow for dedicated time for pre-test counseling within clinic appointments. The integration of GCs outside of clinical genetics departments and MDT meetings may help non-genetics HCPs to consult with genetics specialists about issues regarding pre- and post-test counseling for genetic testing (Adlington et al., 2019), knowledge and information about 22q11DS and rare diseases in general, and follow-up support and referrals.

Participants in the current study reported becoming the experts about their child's condition due to non-genetics HCPs having little understanding of 22q11DS, which is well-documented in wider rare disease research (Budysh et al., 2012; Ward et al., 2022). The findings suggest an association between HCPs' low awareness of 22q11DS with participants' perceived lack of empathy from HCPs. Dissatisfaction due to perceived lack of empathy from HCPs has been reported in previous studies from individuals diagnosed with rare diseases (Zurynski et al., 2017). Carl Rogers described empathy as "the state of empathy, or being empathic, is to perceive the internal frame of reference of another with accuracy and with the emotional components and meanings which pertain thereto as if one were the person" (Rogers, 1980; p140). The findings in this study suggest that for HCPs to display empathy for parents of children

with 22q11DS, they must have an in-depth understanding of the needs and lived experience of the condition. Considering the reported low awareness of 22q11DS among HCPs, it is possible that this contributes to the perceived lack of empathy from HCPs among participants in this study. This is represented in [Figure 2](#).

The Irish Health Service Executive (HSE) has more recently developed an online training module "Rare Disease Information for Healthcare Professionals", resulting from the National Rare Disease Plan for Ireland 2014–2018. Another possible solution could be to develop an online condition-specific training tool for GPs and HCPs working in pediatric and relevant adult healthcare departments, on which that they could self-enroll as necessary.

Participants in the current study report bearing a large responsibility for managing their child's complex care, receiving little or no psychological support and experiencing a lack of understanding of their child's condition among non-genetics HCPs. Findings by Hickey et al. (2020) report that multidisciplinary (MDT) care leads to better outcomes for individuals with 22q11DS. An increased quality of care requires an understanding of the needs of children with 22q11DS, therefore a heightened awareness of 22q11DS may support the provision of effective medical care to this population. The findings from this study suggest a perceived association between HCPs' low awareness of the condition and fragmented medical care, placing a large burden on parents. However, the recent introduction of a specialized 22q clinic has made significant improvements to participants' experiences and improved efficiency by reducing the number of appointments by providing multiple appointments in one clinic. The 22q clinic also alleviates parent's concerns about HCPs with little understanding of 22q11DS, which has previously been reported as a source of frustration for parents in the Irish context (Alugo et al., 2017). However, the reduction of waiting lists and appropriate funding for the 22q clinic is required. An adult 22q clinic may also help to further reduce fragmented medical care experienced by the adults with 22q11DS, where participants describe a decline in the quality of care. This is supported by recent findings by Ward et al. (2022) reporting significant gaps in healthcare services for adults with 22q11DS due to a lack of transition care pathways.

The findings also emphasized a need for children with 22q11DS to be encouraged to develop a good relationship with their GP as they enter their teen years, to help with the transition to adult care and be empowered as young adults. This may tie in with an area for future research to examine the needs of adults with 22q11DS.

A lack of awareness of what genetic counseling entails and of the genetic counseling services available in Ireland by participants and HCPs was identified in this study. This suggests that advocacy of the role of the GC is needed in Ireland, particularly about the GCs' role in supporting families to adapt to genetic diagnoses and to dispel the association of genetic counseling solely with family planning. As some participants in this study may have had unrealistic expectations of genetic counseling, a tailored website or online aid for parents to access prior to their genetic counseling appointment may help to improve expectations of and satisfaction with genetic counseling. This has previously been demonstrated to be useful for individuals attending cancer genetic counseling (Albada et al., 2012).

It is widely acknowledged that families receiving diagnoses of rare diseases for their children should have timely access to genetic counseling (Greenhalgh et al., 2003; Zurynski et al., 2017). Genetic counseling is considered to be a core requirement for care pathways for rare diseases in Ireland (Ward et al., 2022). Ireland has a particularly small number of GCs in comparison to European countries of similar population (Abacan et al., 2019), which hinders timely access for families. By the time parents in the study (those who received genetic counseling) were seen by a GC, they had already educated themselves on the condition and were no longer in a state of shock and the opportunity for GC support and information at the most opportune time had been missed. Addressing the psychological factors and emotions associated with receiving genetic diagnoses, such as blame, guilt, and grief, earlier in the diagnostic journey could support better adjustment and adaptation to the child's diagnosis. The role of the GC is also likely to be helpful for relatives of individuals with 22q11DS to provide accurate information and psychological support (Okashah et al., 2015), as exemplified by the case of a sibling who has decided not to pursue having a family based on their experience of having a sibling with 22q11DS.

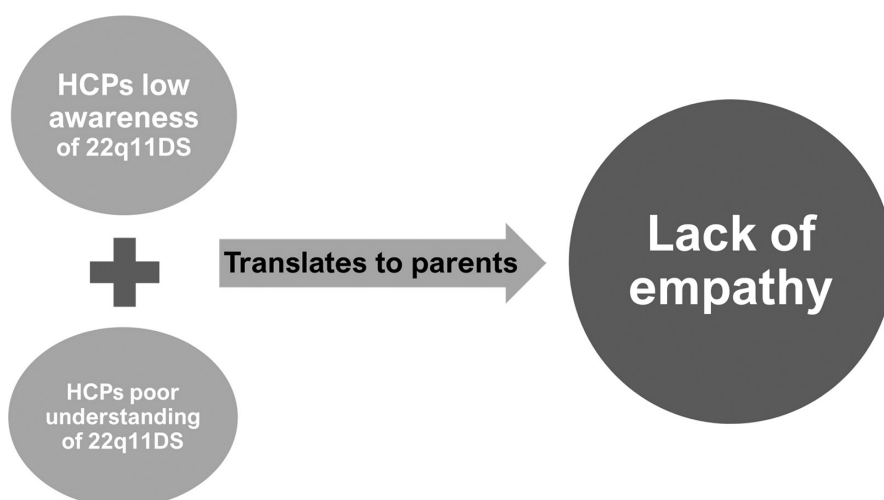


FIGURE 2 Factors contributing to lack of empathy experienced by parents

The delay in access to genetic counseling means that genetic counseling services in Ireland may not be utilized in an effective way by these families. Early and timely access to genetic counseling is also likely to improve health outcomes for children diagnosed with 22q11DS as a result of parents being appropriately informed about the condition, with earlier referrals, such as to the 22q clinic and early intervention services. The timing of genetic counseling shortly after diagnoses is important, with participants voicing that access to follow-up genetic counseling would give parents time to process the diagnosis and prepare questions.

Furthermore, psychiatric genetic counseling has particular relevance to 22q11DS and was specifically mentioned by two participants in the study. Recent research has described an 'Awareness to Act' theoretical framework (as seen in Figure 3) for parents of children with 22q11DS which describes psychiatric genetic counseling as an intervention to assist parents in gaining a proactive awareness of their child's MH in relation to 22q11DS (Carrion et al., 2021). Access to psychiatric genetic counseling for parents of children with 22q11DS may alleviate caregiver stress in relation to MH by helping to understand the association between MH and 22q11DS, with the aim to improve parent's acceptance and empowerment, and reduce potential negative impacts of caregiver stress on the development of MH symptoms in children with 22q11DS.

Psychiatric genetic counseling for individuals with mental illness may contribute to improved outcomes by facilitating understanding

and acceptance through the 'Empowering Encounter' theoretical model (Semaka & Austin, 2019), which adolescents and adults with 22q11DS are likely to benefit from. Of interest is the placement of the needs for genetics and psychiatry in the later adolescent years as illustrated in Figure 4 by Habel et al. (2014), which further supports a need for psychiatric genetic counseling for individuals with 22q11DS.

Overall, several findings in the current study focussing on 22q11DS supports recurring themes in rare diseases, such as the lack of sufficient information provision, low awareness of the condition and parents becoming experts and educating HCPs. The lack of awareness and lack of timely access to genetic counseling may be affecting the wider rare disease community in Ireland. The apparent lack of sufficient pre-test counseling may also have wider implications in rare disease settings. Issues which appear to specifically affect families with 22q11DS include the naming of the condition affecting information provision and awareness, and also the association of psychiatric symptoms and age-dependent clinical features.

5.1 | Strengths and limitations of the study

This study was successful in achieving its aims. A key strength of this study was the relatively large sample size for a qualitative study, achieving data saturation. Another strength of the study was the collection

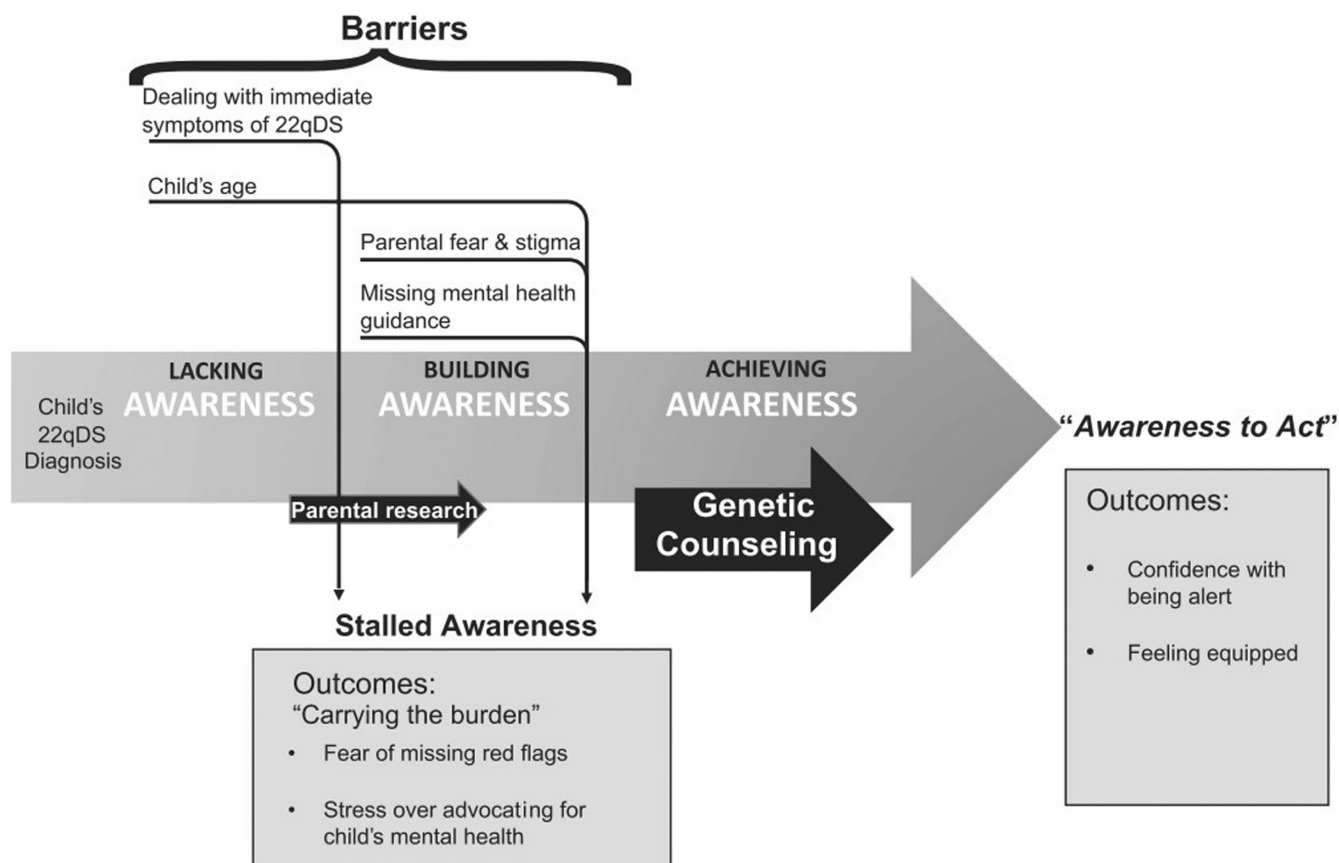


FIGURE 3 'Awareness to act' theoretical framework (Carrion et al., 2021, reprinted with permission)

NEED OF SPECIALTIES BY AGE OF 22q 11 DS CHILDREN

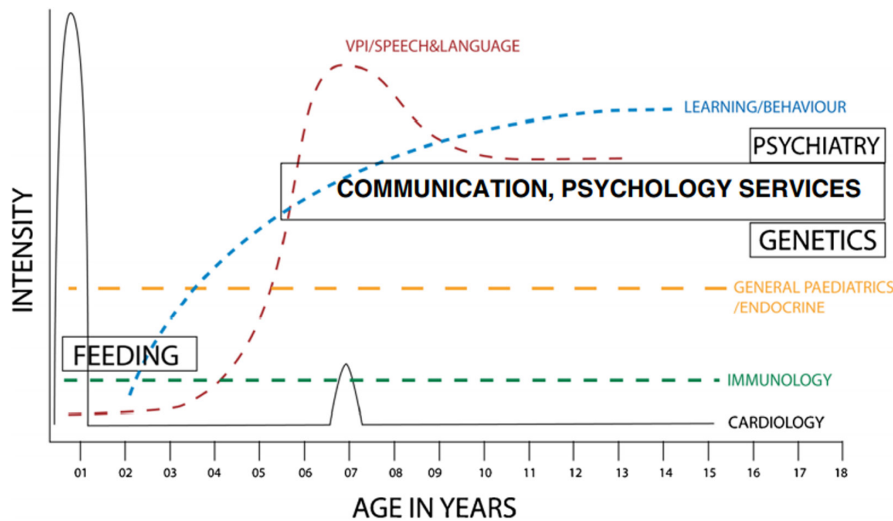


FIGURE 4 Needs of specialties by age (Habel et al., 2014, reprinted with permission)

of data spanning a range of approximately 21 years during which diagnoses were received, the wide range of ages of the child at diagnosis and the different medical specialties that delivered the diagnoses (see Table 1). The validity of the findings of this study is supported by consistent findings identified in previous research (Alugo et al., 2017). The validity of the coding is supported by the double-coding of two interview transcripts by the researcher and project supervisor which showed high concordance. Twenty percent of participants were male which helped to gain perspectives from male caregivers, as much previous qualitative research involving parents of children with 22q11DS heard from the maternal viewpoint as the primary caregiver.

The limitations of the study include the study being retrospective. There is an identified need for prospective, longitudinal research on the impact of 22q11DS on families and individuals. There is a potential inherent sample bias in this type of study where individuals who have had poor experiences with healthcare may engage more strongly with support groups and therefore with opportunities to voice their experiences. Another limitation of this study is that the findings are not transferable to a wider cohort of families due to the small sample size.

The method of conducting interviews remotely may be considered as a limitation of the study. Although this was necessary due to the COVID-19 pandemic, it may have introduced inequity for individuals who do not have access to remote connectivity or a suitable location to attend a remote interview. However, conducting interviews remotely may have enabled some participants to overcome geographic or mobility limitations, thus allowing for the recruitment of some participants who may not have participated if interviews were conducted face-to-face. For those who did participate, remote interviews may have impacted on building rapport, although rapport can be built through other means such as email correspondence prior to interviews (Kessa Roberts et al., 2021). An additional limitation of this study is that only English speakers were eligible to participate, as the research study did not have funding to provide an interpreter for non-English speaking participants.

5.2 | Implications for clinical practice

Some short-term implications for non-genetics clinical practice, include the use of the name '22q11.2 deletion syndrome' instead of 'Di George syndrome' when delivering diagnoses of 22q11DS, along with the provision of supportive information (such as the 'Max Appeal' consensus document, which can be accessed at <https://www.maxappeal.org.uk/consensus-document>) and signposting to support groups. The implementation of dedicated time for sufficient pre-test counseling for genetic testing for 22q11DS may also be helpful; ensuring that parents understand the indications for and potential outcomes of testing and expect to receive results. In Ireland, the implementation of best practice guidelines such as those reported by Harnett (2007) for delivering diagnoses would also be helpful for disclosure of diagnoses of 22q11DS. However, the experiences of the participants in this study highlight the need for adequately resourced clinical services which can feasibly allow for dedicated time for pre-test counseling.

Unmet needs which will require longer-term investment in the Irish setting include the need for timely access to genetic counseling at diagnosis, with follow-up and availability at different life stages, such as at transition to adulthood. Improved care coordination and expanded specialized 22q clinics to reduce waiting lists and relieve parents of high levels of responsibility may also be helpful for families with 22q11DS. Similarly, this may also be helpful for adults with 22q11DS. Due to the lack of support for families receiving diagnoses of 22q11DS, funding for support group activities such as parent and sibling workshops are needed. There is an identified need for non-genetics HCPs to be educated and trained in carrying out and consenting for genetic testing and appropriately delivering results, perhaps through small group sessions and MDT case-based teaching (Humphreys et al., 2021) and the development of best practice guidelines. The further development of online training modules such as "Rare Disease Information for Healthcare Professionals" (can be accessed at <https://www.ucd.ie/vavctest/johntest/NRDO/>)

RD-01%20-%20Storyline%20output/story_html5.html?lms=1) on 22q11DS specifically, and on the psychosocial impact of rare diseases on a family could be utilized for non-genetics HCPs. This study also identifies a need for the advocacy of the GC profession and services in Ireland, in the form of adequate GC staffing levels and adequate resourcing of clinical genetics departments. Potential strategies for increasing the awareness of the GC profession and advocating for patient access to GC services in Ireland could include the provision of education sessions or online training tools for HCPs, university educators, and the lay public; for example, by outlining the role of the GC in different case study examples and the provision of information about how and when to make referrals to genetic counseling services.

5.3 | Implications for future research

Potentially fruitful avenues for future research include exploring with HCPs who are involved in the genetic testing and delivery of results for 22q11DS about their experiences and investigating reasons for the limited referrals made to GCs, as just 6 of 20 participants in this study were seen by a GC. It would also be beneficial to explore whether parents who have received timely access to genetic counseling have better outcomes and experiences of receiving diagnoses and genetic counseling than parents who had delayed access to genetic counseling, or no access at all. There is also a need to conduct longitudinal research with adults living with 22q11DS about their experiences and needs in the Irish context, considering the lack of coordinated adult care identified in this study.

6 | CONCLUSION

The findings in the study suggest that there are several unmet needs for families with 22q11DS, largely regarding the disclosure of diagnoses and the lack of adequate provision of support and information. The findings suggest an association between HCPs' poor understanding of 22q11DS and the perceived lack of empathy from HCPs by participants of the study, and also an association between HCPs' low awareness of 22q11DS and the fragmented care experienced by families.

The study highlights the need for adequate resourcing of GC staffing levels in clinical genetics departments in Ireland, including the advocacy for the role of the GC, and how the current system is not enabling effective, timely access to genetic counseling for families. Parents desire to be proactive with regards to their child's MH, which ties in 'anticipatory care' models (Milic et al., 2021; Schneider et al., 2014). The relevance of psychiatric genetic counseling for 22q11DS has been highlighted in this study, which adds to the need for timely access to GCs for these families. The impact of a 22q11DS diagnosis on the family unit is an area of need in which input from GCs would be warranted.

This study suggests that an increased awareness of 22q11DS among HCPs could help to achieve coordinated care and ultimately lead to better outcomes for these families. The movement toward specialized clinics is also likely a cost-effective strategy for the health service, to reduce multiple waiting lists and fragmented services. The findings support the development of a coordinated care pathway for 22q11DS with stronger input from and timely access to genetic counseling, which may serve as a model of care for other rare diseases.

AUTHOR CONTRIBUTIONS

EOD, MM, and RR confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All 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

The authors EOD, MM, and RR declare that they have no conflicts of interest.

DATA AVAILABILITY STATEMENT

Research data are not shared so as to protect the anonymity of the participants.

HUMAN STUDIES AND INFORMED CONSENT

Ethics approval to conduct this qualitative research was obtained by the Cardiff University School of Medicine Research Ethics Committee in August 2020 (reference SMREC 20/76). Informed consent was obtained from all participants for being included in the study.

ANIMAL STUDIES

No non-human animal studies were carried out by the authors of this article.

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REFERENCES

- Abacan, M., Alsubaie, L., Barlow-Stewart, K., Caanen, B., Cordier, C., Courtney, E., Davoine, E., Edwards, J., Elackatt, N. J., Gardiner, K., Guan, Y., Huang, L. H., Malmgren, C. I., Kejriwal, S., Kim, H.

- J., Lambert, D., Lantigua-Cruz, P. A., Lee, J. M. H., Lodahl, M., ... Wicklund, C. (2019). The global state of the genetic counseling profession. *European Journal of Human Genetics*, 27(2), 183–197. <https://doi.org/10.1038/s41431-018-0252-x>
- Adlington, K., Smith, J., Crabtree, J., Win, S., Rennie, J., Khodatars, K., Rosser, E., & Hall, I. (2019). Improving access to genetic testing for adults with intellectual disability: A literature review and lessons from a quality improvement project in East London. *American Journal of Medical Genetics. Part B: Neuropsychiatric Genetics*, 180(8), 566–575. <https://doi.org/10.1002/ajmg.b.32732>
- Albada, A., van Dulmen, S., Lindhout, D., Bensing, J. M., & Ausems, M. G. E. M. (2012). A pre-visit tailored website enhances counselees' realistic expectations and knowledge and fulfils information needs for breast cancer genetic counselling. *Familial Cancer*, 11(1), 85–95. <https://doi.org/10.1007/s10689-011-9479-1>
- Alugo, T., Malone, H., Sheehan, A., Coyne, I., Lawlor, A., & McNicholas, F. (2017). Development of a 22q11DS psycho-educational programme: Exploration of the views, concerns and educational needs of parents caring for children or adolescents with 22q11DS in relation to mental health issues. *Child: Care, Health and Development*, 43(4), 527–535. <https://doi.org/10.1111/cch.12457>
- Austin, J., Semaka, A., & Hadjipavlou, G. (2014). Conceptualizing genetic counseling as psychotherapy in the era of genomic medicine. *Journal of Genetic Counseling*, 23(6), 903–909. <https://doi.org/10.1007/s10897-014-9728-1>
- Bowen, M. (1978). *Family therapy in clinical practice*. Jason Aronson.
- Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, 3(2), 77–101. <https://doi.org/10.1191/1478088706qp063oa>
- Broley, S. (2013). Acceptance and commitment therapy in genetic counselling: A case study of recurrent worry. *Journal of Genetic Counseling*, 22(3), 296–302. <https://doi.org/10.1007/s10897-012-9558-y>
- Budych, K., Helms, T. M., & Schultz, C. (2012). How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient–physician interaction. *Health Policy*, 105(2), 154–164. <https://doi.org/10.1016/j.healthpol.2012.02.018>
- Campbell, I. M., Sheppard, S. E., Crowley, T. B., McGinn, D. E., Bailey, A., McGinn, M. J., Unolt, M., Homans, J. F., Chen, E. Y., Salmons, H. I., Gaynor, J. W., Goldmuntz, E., Jackson, O. A., Katz, L. E., Mascarenhas, M. R., Deeney, V. F. X., Castelein, R. M., Zur, K. B., Elden, L., ... McDonald-McGinn, D. M. (2018). What's new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. *American Journal of Medical Genetics. Part A*, 176(10), 2058–2069. <https://doi.org/10.1002/ajmg.a.40637>
- Carrion, P., Semaka, A., Batallones, R., Slomp, C., Morris, E., Inglis, A., Moretti, M., & Austin, J. (2021). Reflections of parents of children with 22q11.2 deletion syndrome on the experience of receiving psychiatric genetic counseling: 'Awareness to Act'. *Journal of Genetic Counseling*, 31, 140–152. <https://doi.org/10.1002/jgc4.1460>
- Corvin, A. (2011). Two patients walk into a clinic...a genomics perspective on the future of schizophrenia. *BMC Biology*, 9(1), 77. <https://doi.org/10.1186/1741-7007-9-77>
- Cuthbert, A., Challenger, A., Hall, J., & van den Bree, M. (2019). Information and genetic counselling for psychiatric risks in children with rare disorders. <https://doi.org/10.1101/542423>
- Driscoll, D. A., Salvin, J., Sellinger, B., Budarf, M. L., McDonald-McGinn, D. M., Zackai, E. H., & Emanuel, B. S. (1993). Prevalence of 22q11 microdeletions in DiGeorge and velocardiofacial syndromes: Implications for genetic counselling and prenatal diagnosis. *Journal of Medical Genetics*, 30(10), 813–817. <https://doi.org/10.1136/jmg.30.10.813>
- Du Montcel, S., Mendizabai, H., Aymé, S., Lévy, A., & Philip, N. (1996). Prevalence of 22q11 microdeletion. *Journal of Medical Genetics*, 33(8), 719. <https://doi.org/10.1136/jmg.33.8.719>
- Greenhalgh, K. L., Aligianis, I. A., Bromilow, G., Cox, H., Hill, C., Stait, Y., Leech, B. J., Lunt, P. W., & Ellis, M. (2003). 22q11 deletion: A multisystem disorder requiring multidisciplinary input. *Archives of Disease in Childhood*, 88(6), 523–524. <https://doi.org/10.1136/adc.88.6.523>
- Habel, A., Herriot, R., Kumararatne, D., Allgrove, J., Baker, K., Baxendale, H., Bu'Lock, F., Firth, H., Gennery, A., Holland, A., Illingworth, C., Mercer, N., Pannebakker, M., Parry, A., Roberts, A., & Tsai-Goodman, B. (2014). Towards a safety net for management of 22q11.2 deletion syndrome: Guidelines for our times. *European Journal of Pediatrics*, 173(6), 757–765. <https://doi.org/10.1007/s00431-013-2240-z>
- Harnett, A. (2007). *Informing families of their child's disability: National best practice guidelines*. National Federation of Voluntary Bodies Providing Services to People with Intellectual Disability.
- Hercher, L., & Bruenner, G. (2008). Living with a child at risk for psychotic illness: The experience of parents coping with 22q11 deletion syndrome: An exploratory study. *American Journal of Medical Genetics, Part A*, 146(18), 2355–2360. <https://doi.org/10.1002/ajmg.a.32466>
- Hickey, S. E., Kellogg, B., O'Brien, M., Hall, C., Kirschner, R. E., Santoro, S. L., Leonard, H., & Baylis, A. L. (2020). Impact of interdisciplinary team care for children with 22q11.2 deletion syndrome. *The Cleft Palate-Craniofacial Journal*, 57(12), 1362–1369. <https://doi.org/10.1177/1055665620947985>
- Hill, C., Knox, S., & Thompson, B. (2005). Consensual qualitative research: An update. *Journal of Counseling Psychology*, 52(2), 196–205. <https://doi.org/10.1037/0022-0167.52.2.196>
- Hiller, A., & Vears, D. (2016). Reflexivity and the clinician-researcher: Managing participant misconceptions. *Qualitative Research Journal*, 16, 13–25. <https://doi.org/10.1108/QRJ-11-2014-0065>
- Humphreys, H., Stevens, N., Burke, L., Sheehan, M., Glavey, S., Keogan, M., Rasheed, E., & Faculty of Pathology, the Royal College of Physicians in Ireland. (2021). Core curriculum in pathology for future Irish medical students. *Irish Journal of Medical Science*, 191, 1799–1807. <https://doi.org/10.1007/s11845-021-02774-1>
- Kessa Roberts, J., Pavlakis, A. E., & Richards, M. P. (2021). It's more complicated than it seems: Virtual qualitative research in the COVID-19 era. *International Journal of Qualitative Methods*, 20(1), 1–13. <https://doi.org/10.1177/16094069211002959>
- Lawlor, A., Kerin, L., Orr, D., Leahy, R., Crotty, F., Kelleher, S., Lynch, S. A., Duggan, L., Molloy, E., Altman, E., O'Dwyer, A., Cotter, C., Ward, A., Mc Nicholas, F., & Mc Nicholas, F. (2017). Developing integrated care in the context of rare chromosomal conditions: 22q11 deletion syndrome; a parent/clinician collaboration. *International Journal of Integrated Care*, 17(5), A215. [doi:10.5334/ijic.3525](https://doi.org/10.5334/ijic.3525)
- Liu, Y., Fallet, S., & Koppel, R. (2000). Patients' age at time of testing for chromosome 22q11 microdeletions: Missed opportunities for genetic counseling. *Pediatric Cardiology*, 21(2), 183. <https://doi.org/10.1007/s002469910034>
- MacFarlane, I., Veach, P. M., & LeRoy, B. S. (2014). *Genetic counseling research: A practical guide*. Oxford University Press.
- Martin, N., Mikhaelian, M., Cytrynbaum, C., Shuman, C., Chitayat, D., Weksberg, R., & Bassett, A. (2012). 22q11.2 Deletion syndrome: Attitudes towards disclosing the risk of psychiatric illness. *Journal of Genetic Counseling*, 21(6), 825–834. <https://doi.org/10.1007/s10897-012-9517-7>
- Milic, B., Feller, C., Schneider, M., Debbané, M., & Loeffler-Stastka, H. (2021). Social cognition in individuals with 22q11.2 deletion syndrome and its link with psychopathology and social outcomes: A review. *BMC Psychiatry*, 21(1), 130. <https://doi.org/10.1186/s12888-020-02975-5>
- National Treatment Purchase Fund. (2021). *National waiting list data*. <https://www.ntpf.ie/home/pdf//2021/04/nationalnumbers/outpatient/National02.pdf>
- Okashah, R., Schoch, K., Hooper, S. R., Shashi, V., & Callanan, N. (2015). (2015). Parental communication and experiences and knowledge

- of adolescent siblings of children with 22q11.2 deletion syndrome. *Journal of Genetic Counseling*, 24, 752–759. <https://doi.org/10.1007/s10897-014-9806-4>
- Palmer, E. E., Peters, G. B., & Mowat, D. (2012). Chromosome microarray in Australia: A guide for paediatricians: Chromosome microarray. *Journal of Paediatrics and Child Health*, 48(2), E59–E67. <https://doi.org/10.1111/j.1440-1754.2011.02081.x>
- Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: Results of an online survey. *BMC Family Practice*, 17, 88. <https://doi.org/10.1186/s12875-016-0488-x>
- Reilly, C., Murtagh, L., & Senior, J. (2015). The impact on the family of four neurogenetic syndromes: A comparative study of parental views. *Journal of Genetic Counseling*, 24(5), 851–861. <https://doi.org/10.1007/s10897-015-9820-1>
- Rizzo, R., Van den Bree, M., Challenger, A., Cuthbert, A., Ayllon, M. A., Clarke, A., & Thompson, R. (2020). Co-creating a knowledge base in the “22q11.2 deletion syndrome” community. *Journal of Community Genetics*, 11(1), 101–111. <https://doi.org/10.1007/s12687-019-00425-8>
- Rogers, C. (1980). *A way of being*. Cengage Learning, Inc.
- Schneider, M., Debbané, M., Bassett, A. S., Chow, E. W. C., Fung, W. L. A., van den Bree, M., Owen, M., Murphy, K. C., Niarchou, M., Kates, W. R., Antshel, K. M., Fremont, W., McDonald-McGinn, D. M., Gur, R. E., Zackai, E. H., Vorstman, J., Duijff, S. N., Klaassen, P. W., Swillen, A., ... Eliez, S. (2014). Psychiatric disorders from childhood to adulthood in 22q11.2 deletion syndrome: Results from the International Consortium on brain and behavior in 22q11.2 Deletion Syndrome. *The American Journal of Psychiatry*, 171(6), 627–639. <https://doi.org/10.1176/appi.ajp.2013.13070864>
- Semaka, A., & Austin, J. (2019). Patient perspectives on the process and outcomes of psychiatric genetic counseling: An “Empowering encounter”. *Journal of Genetic Counseling*, 28(4), 856–868. <https://doi.org/10.1002/jgc4.1128>
- Shprintzen, R. J. (2008). Velo-cardio-facial syndrome: 30 years of study. *Developmental Disabilities Research Reviews*, 14(1), 3–10. <https://doi.org/10.1002/ddrr.2>
- Vandeborne, L., van Overbeeke, E., Doms, M., De Beyley, B., & Huys, I. (2019). Information needs of physicians regarding the diagnosis of rare diseases: A questionnaire-based study in Belgium. *Orphanet Journal of Rare Diseases*, 14(1), 99. <https://doi.org/10.1186/s13023-019-1075-8>
- Ward, A., Murphy, D., Marron, R., McGrath, V., Bolz-Johnson, M., Cullen, W., Daly, A., Hardiman, O., Lawlor, A., Lynch, S. A., MacLachlan, M., McBrien, J., Ni Bhriain, S., O'Byrne, J. J., O'Connell, S. M., Turner, J., & Treacy, E. P. (2022). Designing rare disease pathways in the Republic of Ireland: A co-operative model. *Orphanet Journal of Rare Diseases*, 17, 162. <https://doi.org/10.1186/s13023-022-02309-6>
- Zurynski, Y., Deverell, M., Dalkeith, T., Johnson, S., Christodoulou, J., Leonard, H., & Elliott, E. J. (2017). Australian children living with rare diseases: Experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet Journal of Rare Diseases*, 12(1), 68. <https://doi.org/10.1186/s13023-017-0622-4>

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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