

“How can we help our children if no one’s helping us?”: Parent and clinician priorities for mental health support in children with rare genetic conditions who have intellectual and developmental disabilities – a framework analysis

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Abstract

Background Next-generation sequencing technologies are now routinely used in the NHS to identify rare genetic conditions (RGCs) in children with intellectual and developmental disabilities (IDDs). As such, the proportion of children diagnosed in early childhood (<6 years) is rising rapidly. The risk of mental health conditions in this group is high, however anecdotal accounts suggest little mental health support is provided. The present study aimed to (1) define the challenges and facilitators in accessing mental health support for families caring for children with RGC and IDD, and (2) explore family and healthcare professional priorities for mental health support. Focus groups were conducted with parents of children with RGC and IDD and neurodisability medical professionals. De-identified transcripts of interviews were analysed using Framework Analysis. Eleven parents of children with RGC and IDD (anxiety problems $n=7$; behavioural difficulties $n=4$), and four professionals participated in the focus groups.

Results The framework analysis identified general healthcare challenges related to having an RGC, including an absence of information in ‘the big black hole’ of the post-diagnostic void, ‘falling between the cracks’ and being stuck in a loop of referrals. It also identified barriers that were specific to accessing mental health support, including difficulties in understanding how to navigate services, diagnostic overshadowing (where the RGC diagnosis acted as an exclusion criterion for accessing services) and the emotional burden of seeking mental health support. Facilitators in accessing mental health support included parental resilience, happenstance, finding key advocates and getting other diagnoses that were more widely recognised. Parent and practitioner priorities for support were aligned. They included support for families post-genetic diagnosis, training and education for healthcare professionals, early mental health support and care-co-ordination.

Conclusion Children with IDD and RGC diagnosed in early childhood and their caregivers face unique challenges in accessing mental health support for their child. Innovative approaches are needed to support parents

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caring for an IDD child diagnosed with RGC in early infancy and to provide better training for professionals on RGC management.

Introduction

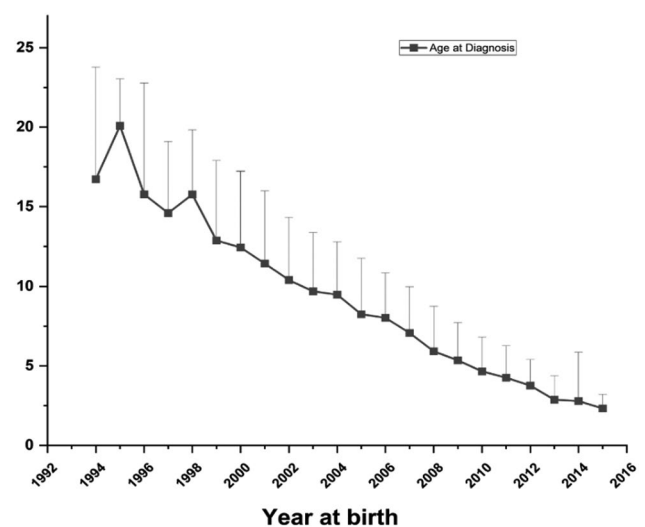
A genetic condition is considered rare when it affects fewer than 1 in 2,000 individuals [1]. Although individually rare, collectively rare genetic conditions (RGCs) are common. In the UK alone 3.5 million people are thought to be affected by rare diseases [2]. RGC are likely to be associated with intellectual or developmental disabilities (IDDs), as up to 84% of genes are expressed to some degree in the brain [3]. IDDs are characterised by significant limitations in cognitive functioning and adaptive behaviour [4]. The aetiology of IDD is broadly divided into environmental exposures (exposure to toxin/infectious, physical trauma) and genetic causes. When the IDD is suspected to have a genetic aetiology, the RGC can be identified in approximately half of cases [5, 6].

The proportion of children identified with IDD associated with an RGC has been rising rapidly since the integration of whole genomic sequencing into routine NHS patient care [7]. Children who are suspected to have developmental delay in early infancy are routinely referred for genetic testing in England, and the age at which an RGC is identified has been steadily declining (Fig. 1). This means there is an ever-increasing pool of infants and children diagnosed with RGC. However, prognostic information on the implications of the individual RGC is lagging behind.

The prevalence of mental health conditions in children with IDD associated with an RGC is known to be high. A large study of children of IDD of known genetic aetiology, including over 1000 different genetic loci, found that 53% met the criteria for a mental health condition [8]. The prevalence rates of mental health conditions of those with both IDD and RGC can vary substantially depending on the individual condition being assessed, with some genetic condition specific estimates ranging from 32% to 74% [9, 10].

The early identification of RGC can be used to guide care, treatment and support. In more common genetic conditions where the physical and mental health phenotype is well described - such as Down's Syndrome or Fragile X - clinical consensus guidelines advise professionals about the most appropriate treatments and share best practice for clinical care with the aim of improving outcomes [11, 12]. Furthermore, there are good examples of RGC-specific treatments for physical health concerns, particularly to optimise the pharmacological treatment of seizures [13, 14]. However, the clinical knowledge base for RGC-specific psychiatric management is sparse [15]. Despite the high risk of mental health difficulties in RGCs, no prognostic information exists [16].

Fig. 1 Declining age of receiving a genetic diagnosis by year of birth in IMAGINE-ID study ($n=2689$) the IMAGINE-ID (Intellectual disability and mental health: assessing the genomic impact on neurodevelopment) programme of research recruited a large national cohort of children with RGC and IDD (Wolstencroft et al., 2022). Their data on the age of genetic diagnosis by year of birth shows a steep reduction in the age of diagnosis over time. In 2015 IMAGINE-ID children received a RGC diagnosis at 2.3 years on average (SD 0.9), compared to 8.3 years (SD 3.5) in 2005 and 20.1 years (SD 3.0) in 1995



At present, both neurotypical and RGC children experience difficulties in accessing mental health support and psychological interventions. The challenges faced by neurotypical children are well-documented [17], but there is little to no evidence on the challenges in accessing mental health support for children with IDD and RGC. A recent report by Spencer-Tansley and collaborators [18], included the voices of people living with or caring for a loved one with a rare condition (including non-genetic and non-IDD participants). It concluded that the drivers of mental health problems were associated with managing a rare condition. They faced specific challenges in accessing mental health support because of the limited knowledge of healthcare professionals about rare conditions. It is important to understand whether there are additional unique barriers or facilitators to accessing mental health support for children with RGC and IDD, to design solutions to address these challenges and optimise opportunities for early intervention.

To date, most of the literature on the psychological impact of having an RGC IDD has focussed on the journey to diagnosis, the so called ‘diagnostic odyssey’. This period of on-going clinical investigations and frequent testing has been described by parents as a time of heightened uncertainty, worry and stress [19, 20]. More research is needed to better understand the experience of parents whose child is diagnosed with an RGC in early childhood and experiences of difficulties with their mental health [21], and the experiences of the neurodisability professionals responsible for their care. The present study aimed to [1] define the barriers and facilitators in accessing mental health support for families caring for RGC IDD children and [2] explore family and healthcare professional priorities for mental health support.

Methods

Study design

A series of focus groups was conducted with a range of stakeholders, including parents of children with RGC and/or IDD, medical professionals working in rare genetic disorders and/or neurodisability, and representatives from UNIQUE, the rare chromosome and gene disorders charity (Fig. 2). Parents of children with IDD and RGC were invited to take part in two focus groups ($n=11$). Separate groups were conducted for the parents of children with

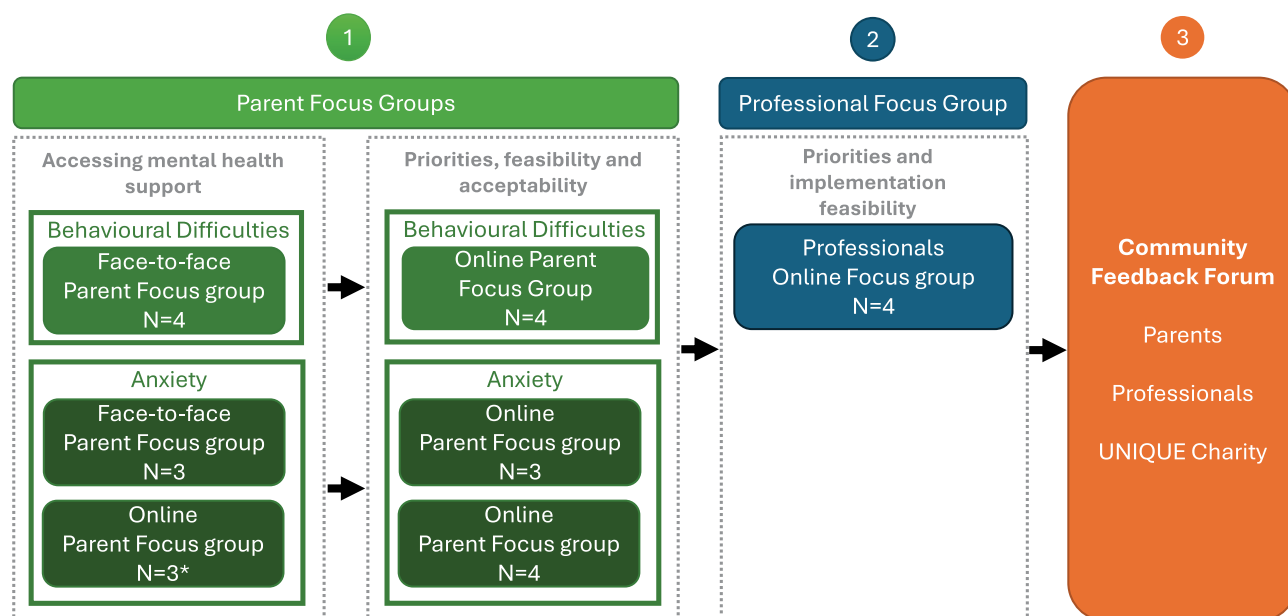


Fig. 2 Focus group and feedback forum study structure. *one parent unable to join the online anxiety focus groups

anxiety difficulties ($n=7$) and for the parents of children with behavioural difficulties ($n=4$). Professionals ($n=4$) were invited to attend one focus group. Charity stakeholders UNIQUE and all focus group participants were invited to take part in the feedback forum. All were audio-recorded and subsequently transcribed for analysis. The focus groups were conducted face-to-face or online and lasted 1h30 each.

Analysis

De-identified transcripts of interviews were analysed using Framework Analysis. Framework Analysis provides a systematic model for managing and mapping the data in seven steps, these include [1]: transcription [2], transcript familiarization [3], coding [4], identifying a thematic framework [5], indexing/applying the framework [6], charting the data into the framework matrix [7], mapping and interpretation [22].

The analysis was led by JW (PhD; principal investigator) and HH (trainee DClinPsy; research assistant). Themes were reviewed following several detailed discussions with the other members of the research team. SB (Psychology BSc; research assistant) applied the framework following a codebook to ensure consistency. The research team comprises female clinical and research psychologists, whose theoretical stance is grounded in a social model of disability. The study team has experience of working with RGC and neurodevelopment. None of the authors are primary caregivers for individuals with IDD or RGC.

Procedures

Children with IDD and RGCs and their parents were recruited from the IMAGINE-ID Study cohort, who had consented to be contacted about future research using purposive sampling. IMAGINE-ID is an Medical Research Council and Medical Research Foundation programme grant which examined the mental health and neurodevelopment of children and young people with IDD and RGCs.(REF) Parents were approached by email, and a sample of those who were available and whose child was between 10 and 15 years of age and had clinically significant anxiety ($n=7$) or behavioural problems ($n=4$) were recruited. Neurodisability professionals were recruited by word of mouth and from electronic mailing lists including the Paediatric Liaison Network and North Thames Child Development Network. Ethical approval for the study was gained from the UCL Research Ethics Committee (9553/007).

Validated measures

Parents were invited to complete validated measures of their child's behavioural difficulties to determine their suitability to be included in the anxiety of behavioural difficulties groups. The Revised Children's Anxiety and Depression Scale (RCADS) is a reliable and valid measure of symptoms of depression and anxiety in children and adolescents [23]. Clinical threshold scores (normal, borderline, clinical range) can be derived based on comparisons to normative data [23]. The Strengths and Difficulties Questionnaire (SDQ) is a well-validated behavioural screening questionnaire [24]. It includes subscales which measure emotional symptoms, conduct problems, hyperactivity, peer relationship problems and prosocial behaviour. Subscales measuring emotional and behavioural problems can be combined to create a Total Difficulties score. A four-fold classification of clinical risk (close to average, slightly raised, high & very high) has been created based on comparisons to UK community samples [25].

In total, four parent focus groups were planned; this included two focus groups for the parents of children with clinically significant anxiety and two focus groups with clinically significant behavioural difficulties. However, as not all families with anxious children were able to attend face-to-face sessions, an additional 'online only' anxiety group was conducted online (Fig. 2). As a result, six parent focus groups were conducted overall. We also planned one focus group with neurodisability professionals and a community feedback forum (Fig. 2).

Face-to-face workshops took place at UCL in Central London and online workshops were conducted virtually over Zoom. Field notes were taken by the facilitators during and after the focus groups.

Focus groups

At the start of each series of focus groups, the facilitators (JW & HH) introduced themselves and shared the study's aims. The first face-to-face parent focus group sessions discussed their children's emotional and/or behavioural difficulties and how they had changed over time, before discussing the challenges and facilitators in accessing mental health support. The second online parent focus group sessions established families' priorities for mental health and behavioural support and discussed acceptable approaches to digital intervention; this was part of a related but separate research question and will be reported in a separate manuscript.

In the professionals' online focus group, a summary of parent focus group discussions was presented at the beginning of the session. The summary included parental reports of difficulties in accessing mental health services. The professionals were then asked to identify their priorities for support and the feasibility of these approaches being integrated into their clinical practice.

Finally, we held an online feedback forum, where we presented our discussions with the parent focus groups to the professionals and our charity partner UNIQUE (Fig. 2). This process served as an opportunity to feedback on our study's key findings to participants and check the accuracy of the representation of the findings. Further 'member checks' were conducted by sending the framework analysis findings to all participants for feedback.

Patient and public involvement

Patient and public involvement was central to the project. The research objectives were developed in response to our experiences of interacting with families taking part in the IMAGINE-ID research project [26], as well as conversations with collaborators at the UNIQUE rare chromosome disorder charity, who are collaborators in the funding application. Participants of this study have been invited to become part of the project's research steering group in subsequent funding applications.

Results

Situating the sample

Seven parents of children with clinically significant anxiety difficulties and four parents of children with clinically significant behavioural difficulties participated in the focus groups. Of the 11 parents, two were fathers and the remainder were mothers. On average, the children were 12.6 years old ($SD=1.8$) with an average parental-estimated mental age of 9 years ($SD=1.6$). The sex split of children was balanced overall, but there were sex-based trends, with more biological females than males in the anxiety group and no females in the behavioural difficulties group. The Revised Children's Anxiety and Depression Scale and Strengths and Difficulties Questionnaire subscale scores confirmed an excess of emotional difficulties in children in the anxiety group and an excess of conduct difficulties in the behaviour group (Table 1).

The professionals' group was comprised of a Consultant Child and Adolescent Psychiatrist, a Clinical Research Fellow working in RGC, a Consultant in Paediatric Neurodisability, and an RGC Nurse Specialist.

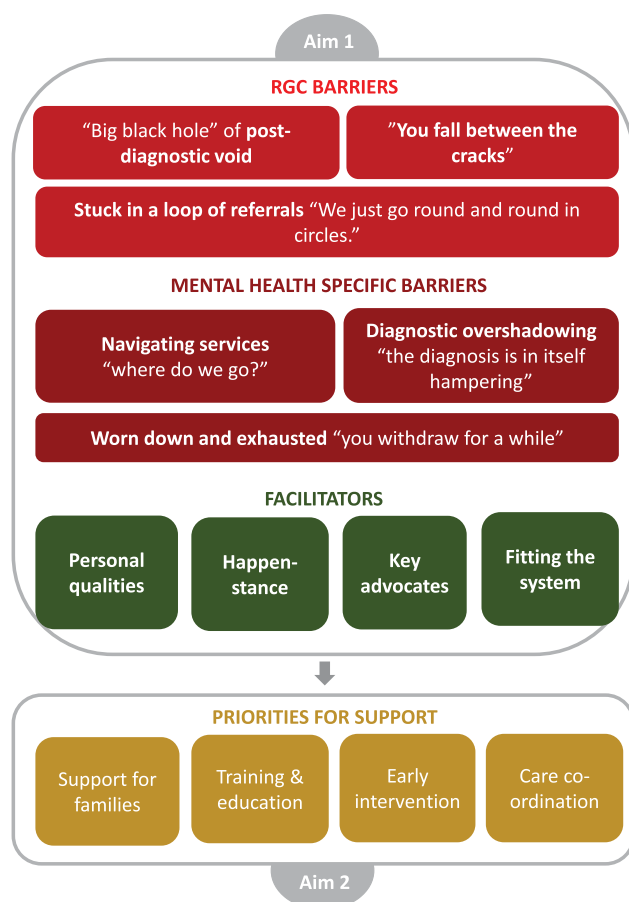
Framework analysis

The framework analysis identified key barriers and facilitators to accessing mental health support for RGC children (Aim 1; Fig. 3) and explored parent and practitioner priorities for support (Aim 2; Fig. 3).

Table 1 Participant characteristics

Child characteristics M (SD)	Anxiety Group N=7	Behaviour Group N=4	All N=11
Chronological Age	13.3 (1.6)	11.5 (1.5)	12.6 (1.8)
Mental Age	9.7 (1.3)	7.8 (1.5)	9.0 (1.6)
Sex (n Male / n Female)	2 male 5 female ¹	4 male	6 male 5 female
RCADS – Anxiety Scaled score	71.5 (10.5) ² Clinical range	61 (11.4) Normal range	67.3 (12.0) ³ Borderline clinical range
SDQ – Emotional problems	8.1 (1.0) Very high	4.0 (3.5) Slightly raised	6.6 (3.1) Very high
SDQ – Conduct problems	2.9 (1.8) Slightly raised	4.3 (1.1) High	3.4 (1.7) Slightly raised
SDQ – Hyperactivity and inattention problems	8.3 (1.6) High	9.8 (0.4) Very high	8.8 (1.5) Very high
SDQ – Peer problems	4.7 (2.8) Very high	4.5 (3.0) Very high	4.6 (3.0) Very high
SDQ - Pro-social ability	6.6 (1.4) Slightly lowered	5.0 (1.2) Very low	6.0 (2.0) Low

¹One female identifies as non-binary; ²n=6; ³n=10

**Fig. 3** Thematic map of framework analysis findings

(Aim 1) Barriers and facilitators in accessing mental health support for families caring for a child with an RGC.

General RGC barriers

The post-genetic diagnostic void

For most families, the lack of information and support started immediately after receiving a genetic diagnosis for their child. Families described receiving the genetic diagnosis like being “thrown into a big black hole”, as little or no information about the genetic condition was available.

Parents described feeling shocked at how little knowledge the professionals had about their child’s RGC: “At that point, and literally, you get the diagnosis, and they say, right, come back in 10 years, and we’ll see whether there’s any other information. So, we didn’t get any sort of help or support at that point”; and how little information they were able to find independently on the internet: “If you get an autism diagnosis, or an ADHD [Attention deficit Hyperactivity Disorder] diagnosis, you can go online, you can Google it. There’s a gazillion support groups and charities and help. But any kind of chromosome differences, whether it’s super rare or a little more common. You’re still very much on your own.”

They described feeling disappointed that there was no provision for follow-up support, aside from being directed to the charity UNIQUE. Some, but not all families were referred to a geneticist for counselling after receiving the RGC for their child: “Our paediatrician gave us a printout: ‘This is what you’ve got, we’ll send a referral letter.’ And that was it.”

“You fall between the cracks”

Parents described that the lack of knowledge and understanding of RGCs was pervasive across the health service: “There’s a lot of these professionals don’t know what a genetic diagnosis means. (...) They’re really uneducated on it.” This meant that parents felt they had to teach the professionals they met about their child’s RGC: “We seem to have to keep educating professionals, GPs, psychiatrists, CAMHS [Child and Adolescent Mental Health Services] on what it all means.”

They described how the lack of understanding of RGCs amongst healthcare professionals led them to feel ignored “I think there’s a lot of professionals who don’t take us seriously, because they don’t understand what the genetic condition is” and dismissed “None of the doctors understand, so it doesn’t get looked into”.

The professionals in our study echoed this experience, and wanted to see their colleagues in other specialities receive some training on managing children with RGCs: “I think that’s something that would be really key for these children is getting that kind of education out there across to specialists, just to get that support.”

“We just go round and round in circles”

Most parents shared a common experience, of being stuck in a loop of referrals for physical and mental health concerns because of the lack of knowledge and clinical pathways for children with RGC: “I get the feeling that they don’t know, so they pass you over. Then we go to the other [professional], who also thinks; ‘I don’t know about that, so I’ll pass you over’. And actually, nothing’s been sorted. Nothing’s helped us (...). My child is super stressed out, because he’s had to see five different people who didn’t have a clue.”

Mental health specific barriers

Navigating services “where do we go?”

Understanding how and where to get help, was a real barrier to seeking support: “We’ve had nothing. I don’t know who to contact. I’ve got no idea.” The professionals reflected that navigating services was particularly difficult for the families of RGC children and young people with moderate learning difficulties. “I think I know that mental health services are really overstretched, and there’s loads of challenges, and I mean the whole NHS is. But particularly that moderate [learning disability] group access to services for that group is really, really difficult, we find, and we just can’t get [them support]. We really, really struggle with that group.” This was often because the young people were considered *too able* for learning disability services, but *not able enough* to engage with services designed for typically developing children.

“Sometimes that diagnosis [RGC] is in itself hampering”

Reports of diagnostic overshadowing were common. This can arise in conditions where there is complex physical and mental health multi-morbidity, when the symptoms of two distinct conditions overlap. Diagnostic overshadowing often results in one condition going undetected or being misdiagnosed. In children with IDD and RGC, diagnostic overshadowing occurred when certain behaviours were assumed to be part of the genetic condition, without exploring other diagnoses: “Once you’ve got a genetic condition, they don’t want to look at the bigger picture and see whether there are any other underlying things going on alongside the genetic condition.”

For example, parents described how it could be difficult to receive a diagnosis of an autism spectrum condition and the impact that this had on the level of support their child could receive in educational settings: “So we didn’t get a diagnosis of autism because we’d had this diagnosis of the genetic condition. This then made it harder to fight when we were going towards an Educational Health Care Plan because the genetic condition wasn’t recognised (...). A lot of problems went with the genetic [diagnosis label].”

The professionals reflected that they had seen many patients who had experienced diagnostic overshadowing and had Child and Adolescent Mental Health Services (CAMHS) referrals turned down because of the RGC: “That diagnosis in itself, is hampering your ability to get this help and support in a way, because of this diagnostic overshadowing. I’ve had letters where they said, well, we can’t assess for autism, because they have a genetic syndrome.”

They also thought that there could be a reluctance from CAMHS colleagues to make neurodevelopmental diagnoses in the presence of an RGC: “We always try to go through CAMHS if it’s the appropriate age for CAMHS and I’ve had letters back, basically saying they’ve got [X genetic condition], so that’s why they’ve got difficulties. They’ve just had this genetic diagnosis, and we don’t want to put another massive, lifelong diagnosis on them.”

For most families, the RGC label acted as an additional barrier to accessing mental health or behavioural support. In our parent focus groups, there was only one family for whom the RGC diagnosis helped the family to receive additional support from CAMHS: “We were fortunate that she has the chromosome abnormality, because that was a way in, that I think we wouldn’t have got had we been trying to battle through.”

Worn down and exhausted

Parents described giving up on seeking mental health support for their child due to exhaustion: “So tired with it all re-telling in the same tales, going through all the same battles while living it for real at home. You know, you do withdraw for a while.” Others described how their battle to access services and support was relentless and endured up until adulthood: “I just get very frustrated and (it’s) very upsetting. It won’t be long before she’s officially an adult, and yet we’re still fighting the fight. She’s not had all this support that she’s clearly needed.”

Most of the professionals recognised the emotional burden of seeking mental health support: “All we hear about is just the long waiting list for those services, and to get advice to get access to those, because the issues have been raised well, like a long time now, but they haven’t been addressed, and parents are usually quite exhausted and angry even at times, and you can see that they are getting so stressed with the situation they are in and they feel like they are not heard. They cannot find shelter.”

Facilitators

Through the experiences of families who had managed to overcome challenges in accessing mental health or behavioural support, key facilitators were identified.

Personal qualities: The first key attribute was the family’s tenacity “You’re thought of as a neurotic parent, because you went back to them saying I’m not happy because we haven’t got any support” and resilience in the face of a challenging healthcare service to navigate: “You’ve got to go through multiple agencies to get to the point where somebody believes you and then actually starts to take it seriously to see what’s actually happening and what kind of support might there be.”

Key advocates: Having a professional advocate helping the family was often a transformative experience in accessing mental health support. For some families the advocate was a staff member at school, for some, it was support from charities and local organisations, and in one instance, it was a medical professional. Most families felt incredibly lucky to have found their advocate and expressed concerns that these encounters had only happened by chance: “One doctor in a hundred gets it”.

Happenstance: Alongside meeting the right people that could help, families also talked about the importance of living in the right area to get help. One family described how moving house had drastically changed the level of mental health input their child had received: “Then we moved from [A] to [B]. [B] are much more open to it. You get so much more support and more help in [B]. We’ve now been to see a CAHMS counsellor, at primary school and at high school.” The professionals focus group were also concerned with the geographical differences in service provision of the NHS: “There’s this massive postcode lottery in terms of mental health support and in terms of the care coordination.”

Fitting the system: The final key facilitator in accessing services was having diagnostic labels that the health, social and educational systems understood: “When we did finally get our diagnosis for autism it was really easy [to access support].” For many families, receiving recognisable neurodevelopmental diagnoses such as autism or ADHD, were critical to accessing further mental health or educational support.

(Aim 2) Priorities for behavioural and mental health support

Parents and clinicians wanted to see better care for RGC children and their priorities for support were remarkably aligned. They all wanted better support and information for families, education for healthcare professionals about RGC, early intervention and improved care coordination (Fig. 2).

Support for families

Parents described wanting better information and support in the post-genetic label diagnostic void. They would have liked support from an expert in the genetic condition just after the genetic diagnosis was made. However, they acknowledged that this was not always possible: “Is there a single person that knows all the different genetic conditions? If only there was.”

In the absence of access to an RGC expert, someone with lived experience “You need someone who has lived it” or “guidance of somebody who’s really in the know” was helpful. Some parents described finding solace in the community, highlighting the importance of peer support: “Parents who ‘get it’ are important. We use some

of those support networks, less so now but certainly early stages of diagnosis. Talking with other parents with youngsters, similar conditions is very, very important, actually, and having that network and support.”

Alongside receiving expert advice about the RGC, they wanted support for themselves: “There’s got to be something for parents as well as the children. When you’re diagnosed with a genetic disorder of any kind, because how can we help our children if no one’s helping us?” Supporting parents through this period of upheaval would enable them to feel better equipped to cope with the additional demands of raising a child with RGC, but also help them to come to terms with receiving a life-changing diagnosis for their child: “You’ve then got to grieve for the child that you didn’t have that you expected to have.”

Training & education for healthcare professionals about RGC

Consistent with the findings for Aim 1, all participants agreed that there was a knowledge and training gap about RGC that needed to be bridged: “It’s almost like this preventative piece of there’s a baseline knowledge within people within the system I think that isn’t there.”

The professionals thought that addressing diagnostic overshadowing in RGC should be a key focus in the RGC training: “Two priorities would be education of genetic conditions, and that having that genetic condition doesn’t mean that you shouldn’t assess and all these other things.” They explained that this would be beneficial for both their healthcare colleagues and families: “I think the message that needs to get across, both to professionals, but also to families, is that diagnosis is helpful. It can direct your thinking.” Education about how common co-occurring conditions are in RGC, would be helpful in planning for effective treatment.

Early intervention

Families thought the RGC label could be used as an early indicator of the need for future mental health and behavioural support, rather than the ‘watch and wait’ approach they’d experienced: “I think it’s almost the obviousness of a chromosome diagnosis, triggering some actions. That seems fairly straightforward.” At present, this occurs for physical health, but not mental health difficulties: “I think, for me in terms of having the genetic diagnosis. NHS were really good focusing on the physical aspects of that diagnosis. So, what could be wrong physically, whether that’s heart, kidneys, brain, all of that jazz, and we had plenty of appointments to look at that. But going forward in terms of mental health (...), there was nothing really.”

The professionals thought access to early low-intensity mental health support could help to prevent difficulties from escalating “You know you may not get full CAMHS for all these children. But there are steps along the way that can still be really helpful, and maybe even prevent them from getting to the point where they need the full CAMHS support.” as well as provide appropriate follow-up care: “They’re never going to be discharged from their care. I think they are going to be a long-term follow-up, because the needs would change as the children grow older.”

Care-co-ordination

Families highlighted the need for help in navigating the healthcare system, in the context of complex physical, behavioural and mental health needs: “It’s difficult when you your face with all these different problems, and there’s nobody there to oversee it all and to just give us some support.”

The professionals echoed that care co-ordination and specialist clinics would be a more appropriate care model for this population, due to the complex “multi-system nature of the condition”. They felt it would help parents to manage what could otherwise feel like a disjointed experience: “I really felt this issue about having continuity of

care was so important, particularly having somebody that liaise with you through. You're negotiating a massive number of different professionals."

It is important to acknowledge that for parents, care co-ordination had a broader focus than the NHS. Good care co-ordination for parents would also involve communication between the healthcare, educational and social care systems. The professionals acknowledged this interpretation, but only commented on potential service changes that were in their remit.

Discussion

Our most important finding was that having an identified RGC diagnosis often acted as a hindrance to accessing mental health or behavioural support. Clinicians were often reported to be ill-equipped to manage their care and/or offer appropriate services for the child's degree of IDD. This resulted in clinicians referring to other specialists, keeping families in a loop of referrals; specialist services such as Child and Adolescent Mental Health Services (CAMHS) or neurodevelopmental assessment services declined referrals based on the presence of an RGC.

Unique barriers to mental health support for families with children with IDD and RGC

Similar to Spencer-Tansley's (2022) findings on the experiences of accessing mental health services for people living with rare disease, many of the difficulties in accessing mental health support were bound-up in the rareness of the genetic condition. But some of the barriers, such as diagnostic overshadowing and navigating services, were unique to accessing mental health support. The genetic diagnosis appeared to be helpful in guiding physical healthcare management, and could also help facilitate access to educational support. This has also been found in other paediatric RGC studies [13]. **But this was not the case for mental health or behavioural management in our study.** One explanation for this phenomenon reported by the clinicians was diagnostic over-shadowing and a reluctance to diagnose additional co-occurring conditions. The latter relies on an implicit assumption that holding multiple diagnoses is a negative, perhaps stigmatising, experience. Another explanation may be the unfounded assumption that behavioural or mental health difficulties associated with an RGC are not amenable to change through intervention, because of the 'genetic' aetiology. There is some evidence to suggest that neurodevelopmental complexity may influence mental health practitioners' decision to reject referrals, reasoning that standard evidence-based interventions are unlikely to be effective and that they require more experience to manage these patients' needs [27]. A competing factor may also be the unprecedented strain under which CAMHS operate, leading services to put into place arbitrary exclusion criteria to manage their waiting lists. Many of the issues described may be ameliorated with better training and education of healthcare professionals. However, a clearer clinical pathway which includes a specification on mental health and behavioural management is also warranted.

Psychological support for caregivers

The consequences of difficulties in accessing behavioural and mental health support are wide-reaching. Childhood mental health problems are often persistent [28] and can lead to problems in adulthood [29]. The potential to contain mental health difficulties, in this high-risk population before they escalate are missed. There is also a huge emotional toll on the caregivers. Parents caring for a child with IDD or a rare disease report elevated rates of mental health difficulties [30–33]. Even parents who had managed to access support for their children felt exhausted - their wellbeing had suffered in the process. Timely psycho-social support for families and clear care pathways or care-coordination could be used to ameliorate this burden. Providing psychological support for caregivers when first receiving the RGC diagnosis has repeatedly been advocated for by the rare disease community [18, 34]. There is evidence to suggest that it can be helpful to reduce stress, burden, and feelings of isolation among caregivers [35].

Support priorities alignment between parents, clinicians and policy

Parent and clinician priorities for support included [1] providing psycho-social support and signposting to families in the genetic post-diagnostic void, where no provision currently exists [2]; educating healthcare professionals on RGCs; and [3] clearer pathways for mental health and behavioural support, early intervention and care co-ordination. Our participants priorities align with the 2002–2025 Rare Disease Framework for England [2, 36] and the Clinical Genomics Service's Accelerating genomic medicine in the NHS 5-year strategy priorities [7]. The Clinical Genomics Service's strategic priorities include the provision of genetic counselling for families, advice for other specialist healthcare providers and genetics education for NHS staff (priorities 1 and 2 [7]). A more explicit focus on providing family mental health care would be needed in future strategic plans to ensure it is not sidelined amongst competing physical health concerns. The Rare Disease Action Plan for England priority areas [36] includes increasing awareness of rare diseases among healthcare professionals (priority 2) and better care co-ordination (priority 3).

The current strategy aims to increase awareness of rare diseases in the nursing and midwifery, pharmacy and primary care workforce, but there is clearly a need to also embed an understanding of genetics in psychiatry and mental health training programmes [15, 37]. The NHS has introduced a genomics notes educational resource for clinicians and healthcare professionals working in the NHS called GeNotes and there have been attempts from the third sector to fill this knowledge gap, with organisations such as Medics4RareDiseases who provide free online courses on rare disease training for medical professionals, and Rare Minds who provide counselling for the rare disease community. However, training and psychological support will need to be embedded in a more systematic way in order to avoid the emergence of new geographical disparities in care provision.

Scoping work on care provision for those with rare diseases (genetic and non-genetic) in the UK has found that care is generally not well co-ordinated [38]. Third sector organisations are piloting care co-ordination approaches such as the UK Same but Different organisation's 'Rare Navigator' service. More innovative approaches are needed to deliver care-coordination in a cost-effective manner across the lifespan for RGC children.

Strengths, limitations and future directions

The study reflected the experiences of the parents and paediatric professionals who were interviewed. Our interviewees were mostly white females; experiences are likely to differ for participants from different backgrounds. Clearly, this small study cannot represent the full extent of the impact of mental health provision in the RGC IDD community ($n=11$ parents and $n=4$ clinicians). This research highlights the unmet mental health support needs of the RGC and IDD community, which have been described in previous research. The strength of our study was to include the voices of multiple stakeholders, including RGC parents and neurodisability clinicians responsible for the children's care.

Conclusion

Children with IDD and RGCs diagnosed in early childhood and their caregivers face unique challenges in accessing mental health and behavioural support for the child. Parent and neurodisability professionals' priorities for support align with those set out by the UK Rare Diseases Framework. Innovative approaches are needed to support parents caring for an IDD child diagnosed with RGC in early infancy and to provide better training for professionals on RGC management.

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Data availability The transcripts generated during the current study are not publicly available to preserve confidentiality, but are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate Ethical approval for the study was gained from the UCL Research Ethics Committee (9553/007). Participants provided informed consent before partaking in research activities.

Consent for publication Participants consented for their data to be include in the manuscript.

Competing interests The authors declare that they have no competing interest.

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