UNIQUE

UNDERSTANDING GENES
& CHROMOSOMES

UNIQUE EXPERIENCES

Living with a rare chromosome or gene disorder

Contents

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Introduction

Unique, The Rare Chromosome and Gene Disorder Support Group, was set up in 1984 by Edna Knight, MBE. In the days before the internet, our original members had a little red book in which they would write about their experiences and then post on to the next family. The Unique community has now grown to over 30,000 members worldwide, with over 10,000 in England. Over 1,000 rare chromosome and gene disorders are represented in the Unique registry. Members are both affected individuals and parents/carers.

There can be significant variation in the impact of having a rare chromosome or gene disorder. They are lifelong genetic conditions which for some people can cause severe learning and physical disabilities, complex medical issues and may be life limiting and life threatening. Others may be more mildly affected or may not be aware they have a condition until they experience difficulties having a family or have an affected child.

Since Unique was founded, knowledge and understanding of genetics and genomics has advanced significantly. New genetic conditions are constantly being discovered, but the rarity of these conditions individually means little is known about them (if anything at all).

Last year Unique celebrated our 40th birthday so we decided to ask what life is like for families and individuals affected by rare chromosome and gene disorders. Over 2,000 people responded to our survey, around 100 attended our focus groups and feedback sessions and over 70 people submitted case studies. We also asked questions on our social media channels and in our private Facebook group.

The responses show that living with a rare chromosome or gene disorder can impact all areas of life including finances, education, employment, family life and relationships. Affected families and individuals often feel anxious, frustrated, stressed and isolated. They can face significant challenges due to lack of information, awareness and understanding about their condition. Accessing appropriate and adequate education and social care support adds additional pressure.

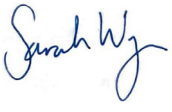
Respondents also told us how determined, passionate and hopeful they feel. How they (or their affected child/adult) are more than just their diagnosis or condition. Many told us how they have become experts by experience in the condition that impacts them and/or their family, and many report how their family relationships and bonds are stronger because of their experiences.

It is now estimated that 1 in 150 babies born in the UK have a rare chromosome or gene disorder. Comparatively, around one in every 1,000 babies is born with Down's syndrome. Public and professional awareness of rare chromosome and gene disorders remains low resulting in affected families and individuals feeling marginalised, isolated and devalued. This must change.

As understanding and advances in genomics continue, equal attention must be paid to the experiences of people and families who are living at the forefront of these developments. It should not be down to 'luck' if a person or family is effectively signposted or empowered to be an equal partner in their care. Greater efforts must also be made to raise awareness of rare chromosome and gene disorders at a local level to reduce the isolation faced by people/individuals and families trying to navigate a system in which they often feel invisible.

The scale of the challenge is significant, but Unique is committed to driving change for everyone impacted by rare chromosome and gene disorders. As our origins from our little red book demonstrate, sharing stories can be powerful. They can create community, promote understanding, evoke empathy and generate action. We invite you therefore to read this report as a repository of stories - each quote, each statistic, each finding is a family or individual's experience. Many of the stories shared with us did not make it into this report but are equally as important as those that did, they all helped us have a greater understanding of the challenges faced. We are grateful to everyone who took the time to share.

Best wishes, Sarah and the Unique team



Executive summary

This report shares the findings from our recent consultation exploring the experiences of families and individuals affected by rare chromosome and gene disorders. The survey received 2,154 completed and 538 partial responses, alongside insights gathered through focus and feedback groups, social media discussions and case studies. The findings reveal a complex picture of resilience, grief, stress, strength and struggle.

Accessing services, lack of respite, uncertainty about the condition, lack of understanding and judgement from others, worries about the future, scarce information, managing symptoms and behaviour, and the complexity of care all impact on family life, affected individuals and their parents/carers.

KEY FINDINGS

1 Lack of awareness and information about rare chromosome and gene disorders creates significant stress.

Lack of professional awareness, low public understanding and struggles to access appropriate information and services create significant challenges for individuals and families affected by rare chromosome and gene disorders. This often results in heightened anxiety, frustration, stress and burnout.

2 Rare chromosome and gene disorders affect every aspect of daily life.

Rare chromosome and gene disorders can affect all areas of life and life chances, including negatively impacting finances, reducing employment and education opportunities, and creating strain on family relationships and fertility journeys.

3 Individuals and families feel isolated.

Having, or caring for someone with a rare chromosome or gene disorder can strengthen close family relationships and bonds. However, living with constant uncertainty, and the complexities of care and managing a condition, often negatively impacts on social networks and relationships resulting in significant feelings of isolation and loneliness.

4 Advocacy is not always valued or a choice.

Many individuals and families become experts in the condition affecting them or their loved one, but their advocacy, insights and lived experience are not always recognised or valued by professionals. This can create additional pressures and responsibilities and can hinder collaborative care.

Unique's commitments:

Over the next five years we will invest in our team to increase our outreach and influencing capacity and develop a strategy to increase the visibility of rare chromosome and gene disorders at a local, national and international level.

- We will develop new information and advocacy resources to empower our members to educate and raise awareness of rare chromosome and gene disorders in their local areas or countries.
- We will also work with our international partners in the genomics community to embed understanding of lived experience of rare chromosome and gene disorders in research and clinical settings.
- We will engage with policy makers to ensure rare chromosome and gene disorders are equally represented alongside more well known conditions such as Down's syndrome and autism spectrum disorders.
- We will run a targeted education and outreach programme in the UK to increase awareness, diversity, inclusion, understanding and representation of rare chromosome and gene disorders within Unique membership but also throughout health, education and social care. We hope the findings can be employed and rolled out around the world.

Methodology and demographics

A combination of approaches was used for our consultation process to provide as many opportunities as possible for individuals and families to share their views in a way that felt most appropriate for them. These approaches included an online survey, virtual focus groups, questions on social media on our public pages and in our private Facebook group and case studies submitted by Unique members. The draft key findings were then also shared at feedback sessions for further discussion with members.

Survey

The Unique survey was open to anyone affected by or caring for someone with a rare chromosome or gene disorder, or their wider family members. It was launched for Rare Chromosome Awareness Day in June 2024 and remained open until the 31st July 2024.

The survey questions were reviewed with members of Unique's Membership Engagement Committee and then shared widely via email, social media and through partner organisations. The questions were a mix of open text, 5-point scales and multiple choice. All consented responses were included in the analysis (including partial responses where the respondent had not completed the full survey).

Survey respondents

2,154 people completed the survey and 538 partial responses were received. All responses were included in the analysis.

Social media

Questions from the survey were also asked in the Unique Facebook group and social media channels to provide additional opportunities to hear the views from those who did not want to complete the full survey.

- 182 comments were received for five questions asked on our public Facebook and Instagram channels.
- 28 comments were received for four questions asked in our private Facebook group.

Focus groups and feedback sessions

We also ran a series of virtual focus groups with UK survey respondents and feedback groups on the initial findings with both UK and international members.

- 45 people attended age-range specific focus groups to share their experiences of parenting/ caring for someone with a rare chromosome or gene disorder.
- 53 people attended the feedback groups to discuss the survey findings and share their experiences.

Focus groups

The focus groups were held over the Autumn of 2024 for survey respondents who had indicated they would be interested in attending follow up interviews or groups to expand on their responses. Groups were split based on the age of the person being cared for. Discussions were informal with prompt questions used as needed to facilitate the conversations.

Feedback groups

Feeback sessions were open to any Unique members to attend. The findings of the survey were presented along with the draft key findings for discussion and comments.

Bias/limitations of the consultation

Consultation methodology

Individuals and families were offered a range of ways to be involved in this consultation, some of them may have participated in more than one way. A degree of technological literacy was required to complete the online survey and take part in the virtual meetings. This may have excluded some people from taking part.

Google translate was used to include survey responses received in different languages. The survey was not however offered in different languages which may have restricted people who do not have English as a first language from responding.

Due to the structure of the survey, we only have ethnicity information for respondents from the UK. This showed respondents were predominantly from a white british background (823 respondents/89%). The findings may not therefore be representative of people and families from a minority ethnic community background. These communities often also experience other intersectional issues such a low socio-economic status and barriers to healthcare that may create additional challenges when managing a rare chromosome or gene disorder.

Demographics of participants

The majority of survey respondents (1731/81%) were mothers/stepmothers caring for someone with a rare chromosome or gene disorder. There were also significantly more mothers attending the focus groups than fathers.

The majority of survey responses (1687/80%) were from families with one affected family member. 1,422 responses were from people caring for someone under the age of 16. Four focus groups were held for parents of young people/adults over 16, with 11 people attending. Six focus groups were held for parents of children under 16 with 30 people attending. The experiences presented in this report therefore may be most reflective of the maternal caring experience, with a focus towards caring for a child under the age of 16.

A small number of survey responses (111/5%) were from individuals affected by rare chromosome and gene disorders. Due to capacity constraints, we did not run any focus groups or feedback sessions during this consultation with affected individuals. We received eight case studies from affected young people/adults and plan further exploration of these experiences in the future, especially those with learning difficulties or disabilities who may not have been able to respond to this survey. We are committed to further consultation with these individuals in the future to ensure their voice is included.

Where survey responses from fathers/stepfathers, affected individuals and parents of adults were significantly different we have presented their responses and views separately from the main data.

Location of participants

61% of respondents live in the UK. The experiences presented in this report, particularly in relation to healthcare and access to services, may not therefore be reflective of experiences of families based in other countries.

- 15% of respondents were located in the USA.
- The other 24% were from various locations including Europe, Canada, New Zealand, Australia, Asia, Africa and South and Central America.

Key challenges

Key challenges faced by families

Survey respondents were asked to tell us in their own words what they thought individuals and families impacted by rare chromosome and gene disorders found challenging.

We asked them to reflect generally on what they felt is the biggest challenge for families affected by rare chromosome and gene disorders and also to reflect how having, or caring for, someone with a rare chromosome or gene disorder has affected them on a more personal level.

Their responses were categorised into themes and these were then further explored in the focus groups.

Difficulties in accessing the right support and services and lack of respite and support were common themes in the survey responses. Stories shared by carers in the focus groups demonstrated how these challenges often lead to frustration, stress and burnout as families try to navigate systems that have little understanding or awareness of rare chromosome or gene disorders. These frustrations are often increased by the financial crises faced by many Local Authorities who are cutting services. Many group participants discussed how difficult it was knowing where to access information about what services were available, or how to access them.

> "Accessing the services their child requires, fighting for what's necessary in their EHCP. Speech and language services on the NHS are particularly minimal and insufficient."
>
> Survey respondent

> "For me, it was getting my child diagnosed in the first place and his needs taken seriously."
>
> Survey respondent

> "Lack of understanding with GPs, in schools, nurseries and in society generally. People look at my grandkids as if they are nasty, naughty kids, but they aren't."
>
> Survey respondent

The key challenges identified for families affected by rare chromosome and gene disorders:

- Accessing the right support/ services
- Lack of respite/support
- Uncertainty about the condition
- Lack of understanding/ judgement
- Worry/uncertainty about the future

ZACHARY

Case study

Zachary has faced more challenges in his young life than most could imagine. He spent his first 58 days in the ICU, sedated and ventilated, and his first 100 days in the hospital. To date, he has undergone three heart surgeries, with a fourth on the horizon, as well as orchiopexy and tympanostomy surgery. Despite these hurdles, Zachary's spirit shines brightly.

He adores singing, puzzles, Toy Story, and trains—his joy and determination are contagious. While developmental delays have been part of his journey, his fierce determination continues to inspire everyone around him.

The greatest challenge for families like Zachary's isn't just the complexity of the condition but navigating broken health systems and advocating for early intervention and support. Rare conditions like KBG syndrome often lack awareness and resources, leaving families like Zachary's in constant fight mode advocating tirelessly for what their child needs. Local supports and functional systems could profoundly change the futures of these children, and charities are a lifeline.

Zachary is no stranger to adversity. When told something isn't possible, he quite simply proves how it is. He's a fighter, a problem-solver, and an inspiration to us all.

Personal challenges

On a more personal level, survey respondents and focus group members shared how challenging they found managing uncertainty (both about the condition and how this created worries for the future) and how they often felt judged and struggled with the lack of understanding of rare chromosome and gene disorders.

Survey respondents also reflected on how a lack of respite and support, accessing the right support and services, and uncertainty about the condition impacted on their wellbeing. Parents/carers also told us about how managing the complexity of care, and the impact of the symptoms and behaviours of the person they cared for felt very challenging.

There was slight variation in what different respondents found personally most challenging. While mothers/stepmothers and fathers/stepfathers both found accessing the right support/services personally challenging, mothers/stepmothers were more likely to personally struggle with lack of understanding and judgment from others and inadequate information about the condition, while fathers/stepfathers felt that lack of respite and the complexity of care/impact on family life was most personally challenging for them.

Personal challenges for affected individuals

The biggest challenges reported by individuals affected by a rare chromosome or gene disorder was the impact and management of their symptoms and behaviours, and lack of understanding and judgement from others. Many respondents also reported explaining their condition and managing the uncertainty around it was difficult.

The impact on their mental health, of living with their rare chromosome or gene disorder, was a common theme for affected individuals. This was echoed in a later question where 44% of affected individuals (37 individuals) reported they did not feel their emotional and mental wellbeing was good. Others also told us they worried about the impact of their condition on their parents.

Other respondents (22/26%) told us their mental wellbeing was good. They told us how their condition or differences did not bother them, or how as they have got older they have learned to manage the challenges associated with it.

Personal challenges for parents/ carers of adults

Uncertainty and worry about the future was identified as one of the key challenges for people caring for their adult child by survey respondents and focus and feedback group participants. In particular, they reported worrying about who will care and advocate for their child when they are no longer able to.

Themes that often appeared in survey responses also included struggles with transitions and poor adult social care support. In the focus groups many participants talked about how difficult it is to plan or know what to expect. They reported there is very little information available for managing new life stages such as puberty, finding post-25 opportunities for employment or meaningful engagement, and independent living arrangements.

Both survey respondents and group participants described how they felt support drops off the older their child becomes. Most were still actively involved in providing and/or managing their adult child's care with some describing particular challenges navigating mental capacity and independent decision making.

The top five personal challenges for all survey respondents most frequently mentioned:

- Lack of respite/support
- Uncertainty about the condition
- Complexity of care/impact on family
- Accessing the right support/services
- Impact of symptoms/behaviours

Most common words chosen by individuals affected by rare chromosome and gene disorders to describe their experiences:

- Anxious
- Frustrated
- Stressed
- Misunderstood
- Depressed
- Lonely

What is the biggest personal challenge for you due to having, or caring for someone, with a rare chromosome or gene disorder?

"Seeing my beautiful daughter suffering and can't do anything."

Survey respondent

"Personally it has been the biggest challenge of my life. Trying to be fully knowledgeable, to have all the resources, to juggle appointments and therapies. To manage home life including other siblings. Isolation."

Survey respondent

"Few understand that the caregivers in my family have jobs that must be kept intact in order to care for the individual; they seem to think we have infinite time and/or resources to handle care - many judge my husband and I for not being able to overcome behavioural hurdles that get in the way of the individual's care."

Survey respondent

"Being understood that I am not a standard bulk issue 46XX yet expected to conform to social norms on every level. 'If you look okay, therefore you are'. No help or concession given".

Survey respondent

"Not many people have heard of chromosome disorders, and knowing my specific one is even harder. I feel so misunderstood and lonely."

Survey respondent

"For me personally I find it so difficult to be accepted, understood and respected as someone with a rare chromosome disorder. I am also autistic as well. I find it difficult to fit in, find work and do anything socially as I don't have friends."

Survey respondent

"My parents don't get support and I am concerned about their mental health."

Survey respondent

"Controlling my emotions and understanding them. Also having the strength to get through the day."

Survey respondent

"Uncertainty about the future. My daughter was diagnosed at 18 months and so her adulthood seemed a very long way away. There was a fair amount of support for her as a baby and young child but the support and services reduced and seemed harder to access as she progressed into her teenage years and into adulthood."

Survey respondent

"The fear of old age and illness which would prevent me caring."

Survey respondent

"There are no ethical counsellors to ask for help on certain issues such as sexual issues, pregnancy, etc. Sexual development is a challenge when our sons are teenagers and social behaviour. In a world full of gender attacks, and claiming for gender diversity respect, our son´s sexual behaviour (with scarce inhibition) can be misunderstood with horrible consequences."

Survey respondent

"Getting a diagnosis took 18 years with no joined up care for all my son's symptoms. Even now, our nearest clinic for [their condition] is only for children. So although there is a body of expertise available, we only have indirect access as he is an adult."

Survey respondent

"Not having freedom in my life. This means I cannot go on family holidays, a cruise, decide what to do and when I am 63 years old, semiretired, and my 33 years old son who lives with us requires 24 hour care."

Survey respondent

RACHEL

Case study

"I WANTED BETTER; FOR MY DAUGHTER TO LIVE AS FULL, INDEPENDENT AND HEALTHY A LIFE AS POSSIBLE"

My daughter is 24 with mosaic trisomy 9. Although often delightful she can be hard work with moderate to severe learning disabilities and occasional violent or sulky outbursts. She is unable to live independently and will always require constant care. For much of her teenage years I was her sole carer. The toll this took on me personally was considerable and has been described as a 'life sentence'. I wanted better; for my daughter to live as full, independent and healthy a life as possible and similarly for me while also moving on from child rearing. The wishes surely of so many mums. The decision for her to move into supported living aged 20 was, and remains for us, sound. But it's not perfect and some were shocked that I considered the option. The standard of care and ambition hasn't aways been good enough although after a second move earlier this year it is now better. I remain closely involved, parenting in a different way, but who will advocate for my daughter when I am no longer able to do it? I love my daughter dearly, speak to and see her regularly but want so much more. And is it wrong for me to want a life of my own too? There may be an element of chance in a child's genes, but should their adult care be so much of a lottery?

KEY FINDING 1

Lack of awareness creates significant stress

Lack of professional awareness, low public understanding and struggles to access appropriate information and services create significant challenges for individuals and families affected by rare chromosome and gene disorders. This often results in heightened anxiety, frustration, stress and burnout.

Professional awareness

47% of survey respondents felt information about their rare chromosome or gene disorder had been hard to access, with many respondents noting that the only information they had been able to access was from Unique.

Some parents/carers mentioned that they found it hard to advocate effectively for their child because they could not access information to help them understand the condition.

In many of the discussions participants expressed how they felt rare chromosome and gene disorders were invisible or ignored because they were not as well known or understood as autism spectrum disorders (ASD) or Down's syndrome. Some families whose child also had a diagnosis of ASD, or autistic traits discussed how this could be useful as it was more widely understood and 'unlocked' access to services, but also that it often meant the rare diagnosis was ignored and sidelined.

> "It's relatively unknown and there is no support .... I wish my kid had Down's syndrome."
>
> Survey respondent

> When A was about 14, she then got a diagnosis of autism, and once she got that diagnosis, people could latch on to that because they could understand it. And so the chromosome issues have sort of kept being pushed to the back and she gets really frustrated by that... because people just want to focus on that one thing that they feel they can cope with."
>
> Feedback session participant

Frustration and stress

Frustrated and stressed were two of the top five words selected by survey respondents to describe their experiences.

Other related words included:

- Powerless
- Disheartened

It was clear from the discussions in the focus groups that managing these feelings had a significant impact on people's wellbeing, many were very honest about how burned out they felt and how this lowered their capacity to cope with the other demands on their time such as managing and/or providing care for their child. They were often very specific in sharing it was not their child who was the cause of their frustration and stress, but rather the systems and services they were having to navigate in order to get them the support they needed.

> "Our experience has been stressful to say the least, trying to come to terms with a rare diagnosis and the uncertainty around it, whilst trying to manage his very immediate medical issues has been extremely hard on us."
>
> Survey respondent

> "People expected me to be relieved that the diagnosis has given me the answers to my child's struggles but I felt frustrated that this could have been diagnosed years ago but I hadn't been listened to and professionals kept disregarding my worries."
>
> Survey respondent

Lack of awareness creates significant stress

Healthcare professionals

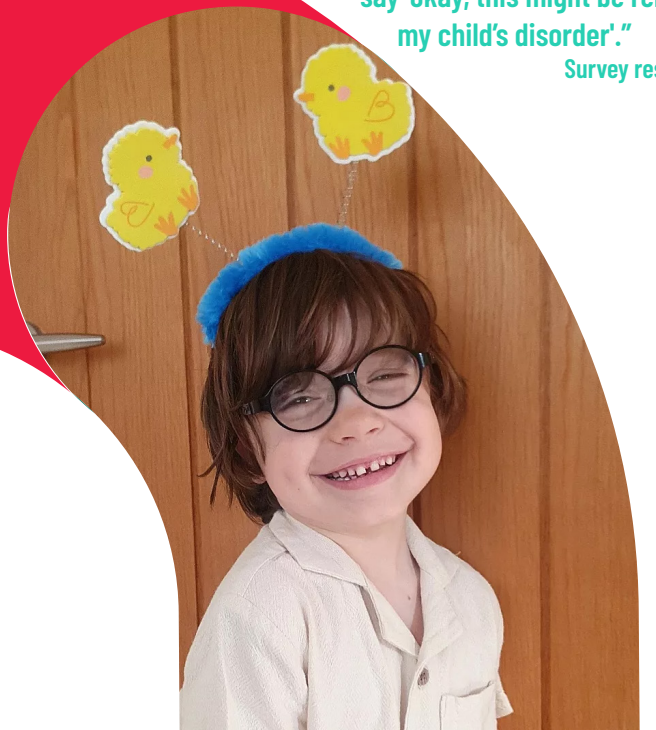
63% of survey respondents felt that they/ their healthcare professionals did not have a good understanding of their rare chromosome or gene disorder.

The impact a well informed versus uninformed healthcare professional could have was a common theme throughout the consultation. Some participants shared stories of how professionals had advocated for them in the early days of diagnosis and how this had helped them access appropriate tests and care. Many others shared how they felt they had to battle for their child's needs to be taken seriously, that they were treated as over anxious parents and felt they had to take responsibility for educating and advocating for their care.

> "Lack of up to date information about rare gene disorders within the medical field. This leads to a lack of support services being made available for your child. There is also a huge lack of awareness of rare gene disorders."
>
> Survey respondent

> "Medical professionals don't know specifics about the deletion my daughter has - just treated as chromosome deletion."
>
> Survey respondent

> "The fact that when you search for anything known about my child's disorder and nothing is found, all your child's doctors know nothing, you have rely on the experiences of the 3 other parents whose children are older than your child to say 'okay, this might be related to my child's disorder'."
>
> Survey respondent



Education professionals

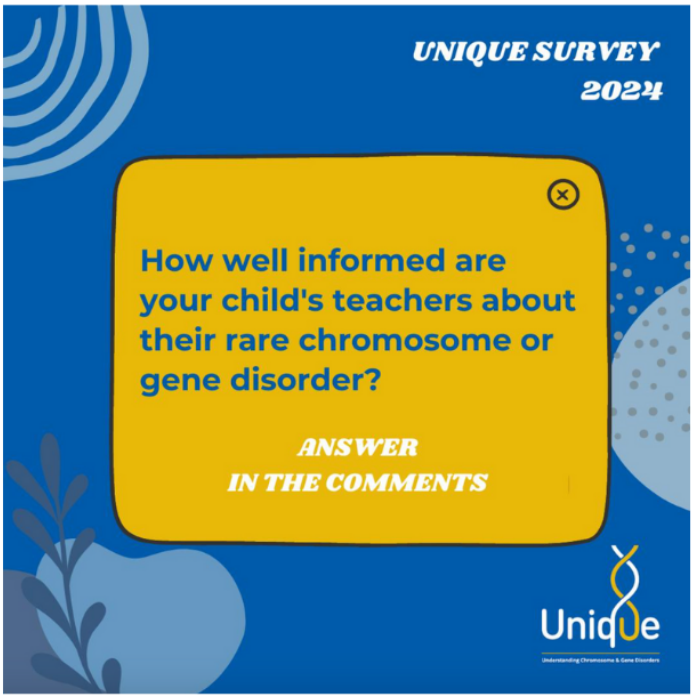
46% of survey respondents felt teachers or educational staff lacked awareness of their rare chromosome or gene disorder.

This was echoed in the focus and feedback groups and on social media responses with many highlighting the positive impact a well-informed (or willing to learn) educational professional could have. Responses suggested that it was often down to the individual teachers to take the initiative and learn about the condition, and that there was not a systematic way that information was provided to them – in most cases it seemed the main source of information was the parent providing the relevant Unique leaflet.

> "I think it's a lottery, a post code lottery again, because when my T was really young, he had a really good preschool nursery where it was excellent and wonderful. Then he went into a special school or as a unit attached to an ordinary primary school and the head teacher there had no understanding at all, and she kept telling him off and punishing him and his behaviour was down to his chromosome abnormality. So you have different professionals who are either helpful or not, and it's a bit of a lottery."
>
> Feedback session participant

> "They don't really know anything about it, they've never mentioned it. I don't even think it's part of her EHCP as the woman handling it all was super snotty and said as it had no clinical significance then it didn't need to be there."
>
> Instagram comment

Lack of awareness creates significant stress



Fully informed. They just cannot make things work for our Unique boy

15w Like Reply

They don't seem to be that interested. I'd love them to know more about neurogenetic development conditions at large.

15w Like Reply

I printed off the Unique Booklet when my son was first diagnosed and gave a copy to Sendco and class teacher.

15w Like Reply

I had the school system put the unique booklet with highlights of what pertains to her in her file and have a meeting every year to discuss it. They are great with it and her!

15w Like Reply

No one in my daughters care is and not interested . That includes health .

15w Like Reply

Not very, they are learning some things . Although my daughter is still a pre school no mainstream will accept her at the moment:(

Meghan Rohrer
I printed off the Unique booklet for BRPF1 & they were so happy to have something that explained his condition a bit more 🫶

15w Like Reply

Not at all. I have been wondering if I should include their information to the teachers

15w Like Reply

No one is as to date there's only three cases documented ,that's myself and my two children ,no one knows anything so I can't really expect a teacher to know if even the experts don't know

15w Like Reply

My school doesn't understand it and think that he is just not interested in learning when in fact it's the complete opposite I wish this was spoken about more and more all I ever hear about is from the is he has adhd and displaying signs of being autistic 🙈🙈🙈🙈 my son was born with deletion 15q and 23

15w Like Reply

They are very aware!! I make them aware and it just happens that I'm a teacher at the school he attends! I make sure every single teacher knows about everything with him. Last year we were right next door to each other and I made sure his teacher knew! Without me telling them they have no idea about any of it. As a teacher I'm very aware and interested In helping parents anyway I can.

15w Like Reply

I gave them the unique booklet and marked the parts that were relevant in relation to our daughter. Her symptoms are rather mild, so it was important for us, that school, and prior to that, daycare didn't Google or read on their own. If they had done that, I firmly believe they wouldn't have deemed it possible for our daughter to attend.
Information is important. However, it needs to be specific for the child or else the relation to the child and how the staff is approaching the child will be biased instead of open minded.

Luckily, most staff has had the same approach. That way the information is dialogue-based. There's overall a big interest in our daughter's needs, and by now a lot of knowledge.

15w Like Reply Edited

Not really that interested. My child's (uk) mainstream school tends to focus on recognising barriers and challenges to learning to the individual. There's advantages and disadvantages to this approach. I'm not sure how it could change even if they included further background research in rare genetic conditions or in my child's specific condition. However, I would want schools to have more access to providing case reports and studies where there are known diagnosis of the genetic conditions. It might then be easier to research more targeted support methods so that genetic conditions could be included in the meta data of the ALN research for more precise results (eg pico searching)

Responses from the Unique Facebook page

Access to information

47% of survey respondents found it difficult to access reliable information about the rare chromosome or gene disorder that affected them or their child.

In the focus groups almost all the participants told similar stories of being given their diagnosis and then 'sent away with a Unique leaflet'. Participants affected by a condition that does not have a Unique leaflet, or where the leaflet had not been updated, reported struggling to find information.

The lack of available information had a profound impact on parents struggling to process the diagnosis and understand what this meant for their child's future. If any information was available, this was often medical or research papers that presented the 'worst case scenario' which was distressing for families to read.

Others also reported how they struggled to understand the information that was available and how this could then impact on their ability to explain to others.

For most respondents and group participants their main source of information was other families with many involved in online groups for their specific rare chromosome or gene disorder. These groups provided valuable community generated information, although this usefulness was limited for parents caring for their adult children. As their children were often among the oldest living with the condition, there was little information available for them about what to expect. They felt very much that they were constantly heading into the unknown creating a pathway for those behind to follow.

These smaller communities were not available for group participants, especially if their condition was ultra rare and/or their child the only known case. For these families information was almost impossible to access.

> It takes a lot of digging and research, usually late hours after your caring or treatment in my instance, making sure my daughter was OK and healthy and once she was napping I was then looking for information, support and answers all hours of the night with little to no information on [her] rare condition."
>
> Instagram comment

> And at the time we were told then that the genetic lab that did the testing said they'd never seen it before. And they've done research and they found that there were seven cases in the world ever at that point. And we were sent medical papers. And it was so scary because there were all sorts of horrendous conditions and in fact, K's very much more, you know, nowhere near as bad as these but it was very scary. All that's all I do remember."
>
> Feedback group participant

> I myself am an NHS medical consultant and found it really hard even with a medical degree, to understand her genome report and the one article that's out there on her condition."
>
> Survey respondent

> I find it very difficult to explain my son's disorder when I haven't been explained to about what it is in detail."
>
> Survey respondent

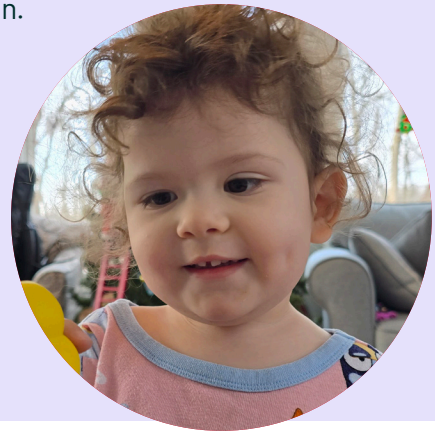
> And also we were referred to a geneticist at one of the hospitals. We had a really positive meeting with her when he was first diagnosed. The geneticist said if we ever needed any help or information at any time in the future, we could contact her at the time, and that she was assigned to him specifically. It felt like we probably wouldn't need to at the time, but since then, with recent changes, I felt we did need to contact her to just see where we go from here. Which hospital/department or consultants she would she recommend relating to his condition? The email address was already resent and only a year later. There was no telephone number in the letters, we felt we were just left in the lurch. When I did find the right email address the reply I received was dismissive and just suggested we seek medical help through the same routes as it he didn't have the chromosome difference, even though the symptoms were ones listed for his condition. So we feel abandoned, I suppose."
>
> Feedback session participant

> There is not a lot of information about chromosome and gene disorders and there are so many variables in each case it is hard to figure things out at times."
>
> Survey respondent

AILA Case study

Aila is currently 2 and a half. From birth she fell behind on milestones quickly, failing to meet any major ones without intervention. We started physical therapy/PT at 8 months and despite it helping her reach milestones, she has never fully caught up to the level of her peers. We saw a neurologist at 18 months when it became clear that something was going on. Her neurologist found minor abnormalities in her brain through MRI and additionally directed us to genetic testing where we found that she was positive for 1q21.1q21.2 microduplication. Upon finding this out we felt so lost and scared. There was little to no information we could find on this condition and her neurologist and paediatrician had little to share with us. We felt very fearful and isolated.

> "UPON FINDING THIS OUT WE FELT SO LOST AND SCARED"



Access to services

64% of survey respondents reported struggling to access social support, respite and short breaks.

Difficulty accessing services was a common theme throughout the survey responses, group discussions and case studies.

It was identified by survey respondents as one of the biggest challenges for families, and many group participants talked about the impact the constant 'battle' for support had on them personally.

A common theme in the group discussions was the lack of support available due to pressures on local authority budgets, and how this was compounded by low levels of awareness and understanding of rare chromosome and gene disorders. Information about what they were entitled to and what was available was difficult to access.

Similar to experiences with education and healthcare, access to services often seemed to be dependent on one engaged professional. Many group participants talked about access to services and information being based on 'luck' and had very low confidence in services systematically meeting their needs.

> I'm sure it's the same across the country. It's because everything is reliant on funding and the government is, you know, pulling the plug on everything now. ... everything has gone recently, they've stopped school transport. They've asked us to make our own arrangements and you know, everything is being cut."
>
> Feedback group participant

> Well, our sort of initial experience was quite different and I think it was just by fluke and luck because we got our diagnosis while we were still in special care. But our consultant who was there when she was born so became her consultant just by accident, had a child with a rare chromosomal disorder himself. So his approach was entirely different and yes, we got the leaflets and things and he sort of signposted to Unique, but he was very much more of an advocate than other people have received and that was just pure luck. But then when he came out of our lives, we just dropped off the edge."
>
> Feedback group participant

> You know you're constantly on, not attack mode, but on defense because you're defending your children, you go into meetings thinking that you're gonna have to fight for something, and then when you realise you don't have to fight for something, you end up bursting into tears."
>
> Feedback group participant

Case study

THOMAS

"NONE OF THIS HAS BEEN STRAIGHTFORWARD"

Thomas was born in 2004 along with his non-identical twin brother Archie. It was clear from the start that Tom struggled a lot more than his brother. Despite all the challenges that he faces, and how difficult he finds everything that most of us take for granted, Tom always rises to the occasion and works tirelessly to achieve his goals. As a toddler, once he started walking he would practice over and over again. Holding hands with his brother he would walk round and round the kitchen table just practising that new skill. Eventually Tom discovered cycling and hasn't looked back. Tom rides regularly with very capable mainstream groups, with a Special Olympics cycling club and competes every year in the Special Olympics GB National Championships where this year he won a gold medal plus 2 silver medals in the top/'elite division' road races and time trials. Tom has completed the Mallorca 225 event (225 km in one day), has completed the RideLondon 100 (100 miles) three times and last year was awarded a Guiness World Record for being the youngest person with an intellectual disability to complete a registered 100 mile bike ride.

None of this has been easy or straightforward. My husband and I had to fight every step of the way to get Tom into the right schools and get the right level of support. Like many parents of SEN children we were pushed to the very brink of tribunal twice, plus have had to fight our Local Authority for Tom's needs for both post 16, continuing post 16 and most recently post 19 placements. This has caused no end of stress, lost sleep worrying about the possible outcomes and has cost us significant amounts of money. Now Tom is older we have found that once in the right environment where he will be offered opportunities and set challenges at just the right level, he will step up to meet those expectations and grow in confidence.

Access to support

Participants were asked whether there were particular times when they most needed support.

Times when families most needed support:

- After diagnosis
- School years
- During medical care or illness
- Pre-diagnosis
- During transitions/new stages

Respondents reported a need for support throughout their lives, from before diagnosis and during medical illness, to major life transitions and into adulthood. The period after diagnosis was identified as the time when support was most needed, followed by support during school years.

Periods of illness and hospital stays were described as stressful and creating additional pressures, including the childcare of other siblings during this time. Times of transition and new stages, such as between schools, or into adult services, were also mentioned as a time when people needed support. Most parents of adults (45/92%) mentioned needing support during adulthood, indicating that the requirement for assistance does not diminish over time, but persists and evolves as circumstances change.

> "After the diagnosis I would have needed help, but I didn't receive it. There were no answers to how it would affect our children's life and also no support to accept the diagnosis. I didn't receive any help at that time. It was: sorry, it's unknown, so it could be everything or nothing."
>
> Survey respondent

Public understanding

As explored more from page 21, lack of understanding and awareness of rare chromosome and gene disorders can often lead to feelings of isolation. Lack of understanding and awareness was identified by affected individuals who responded to the survey as one of the key challenges they faced.

In the group discussions and case studies people also talked about how they sometimes found it hard to ignore people's stares and comments. Some of the group participants also described how sad they felt that their child wasn't valued by society.

Many of the parents/carers described how the constant fight for support and caring for their child affected them mentally and physically, but they had little time to process this and just had to keep going.

> "I've never been able to find a paid job since leaving college 12+ years ago - nobody has time, patience to help me and accept I have struggles and limitations - instead I'm left behind and ignored. People get my hopes up so high and I am always let down."
>
> Survey respondent

Impact on mental wellbeing

The lack of awareness, support and information often leaves individuals and families feeling overwhelmed, emotionally depleted, and alone in managing often complex conditions.

Survey respondents and members of the Unique Facebook group were given a list of words and asked to choose the ones that they felt best described their experiences (they could choose multiple words). They could also add words to the list. The top five words selected were the same for both – anxious, frustrated, stressed, isolated, determined.

Other words added included fatigue, exhausted, proud, guilt, patronised, inferior, fear, tired, privileged, confused, resourceful and overwhelmed.

> "I don't want to blame our daughter in any way. Let me be clear that she is an absolute blessing. But, I've had to make so many sacrifices in order for her to maintain her wellbeing. Mentally and physically I'm exhausted trying to keep her healthy and happy while also trying to tend to my son and spouse."
>
> Survey respondent

KEY FINDING 2

Rare chromosome and gene disorders affect every aspect of daily life

Rare chromosome and gene disorders can affect all areas of life and life chances, including negatively impacting finances, reducing employment and education opportunities, and creating strain on family relationships and fertility journeys.

Finances, employment and education

Parents and carers

65% of parents and carers reported that their finances had been negatively impacted by having or caring for someone with a rare chromosome or gene disorder.

68% said their education or employment had been negatively impacted.

For many this was a direct result of their caring responsibilities and having to give up work completely, or their existing work to try and find flexible part time work.

Many also noted the additional costs associated with having a family member with a rare chromosome or gene disorder – and that for parents caring for their adult children, these additional costs were lifelong.

Group participants also shared how difficult they found it trying to support their children after leaving school to find suitable education or employment opportunities.

> "My relationship with my ex-husband broke down within a matter of months after my daughter was diagnosed with her chromosome abnormality. Trying to find a job with part-time hours as a single parent that fitted in with all her medical appointments was virtually impossible."
>
> Feedback group participant

> "I have to leave my career behind as my son's need became bigger and far more complicated the more he grows with the same severe developmental delays, financially it cost a lot to find care for a disabled child."
>
> Survey respondent

> "Financially, we have been negatively impacted by our daughter in several ways. I was unable to continue my career as a teacher and relied on supply teaching to compensate. We have moved house on 3 occasions to live near our daughter in order to support her whilst living in residential care. We have had to buy her a flat and now have a mortgage (I am 73 and my husband is 75)."
>
> Survey respondent

Affected individuals

67% of affected individuals reported a negative impact on their finances because of their rare chromosome or gene disorder and on their employment and education.

For some this was because they could not access financial support, for others because they could not find suitable or supportive employment opportunities.

> "There's no cure so I feel that life is an endless struggle. I have to work because my disability isn't classed as severe enough to claim benefits. But I'm struggling to work and it just makes life exhausting and pointless."
>
> Survey respondent

JACINTA
Case study

My daughter, Jacinta, is 46 years old and is possibly the oldest person in the world with Kleefstra syndrome, a rare genetic progressive neurological condition. She wasn't diagnosed until she was 41, so we spent years in the wilderness. My older children adore their little sister but there have been many times when we could not have other kids over due to her challenging behaviours or simply because we were too busy managing our special child's needs to be able to go on outings with friends.

My husband and I both closed our businesses when Jacinta developed mental illness at age 24. This was her first 'regression'. She has had three more since then, with the last one starting two years ago. Our family friends have been enjoying overseas travel and pursuing their goals after becoming 'empty nesters'. But, we wouldn't change places for anything in the world.

Family and relationships

Encouragingly survey respondents overall felt that the emotional and mental wellbeing of their family was good (660/41%). Many survey respondents and group participants talked about how having a family member with a rare chromosome or gene disorder had made them stronger as a family.

Family relationships were discussed in the focus groups with many parents sharing how, while their immediate nuclear family unit was stronger, they felt their extended family did not, or could not, accept or understand their child's needs. This often led to them avoiding family events and distancing themselves. Genetic inheritance of a condition in a family was also identified as a potential tension in relationships.

In the groups many parents discussed how they felt their other children had been impacted by having a brother or sister with a rare chromosome or gene disorder. Many felt this had been a positive experience resulting in a strong sibling bond and their children being more caring and supportive, others also recognised the additional pressures this created for them growing up and felt guilty about not being able to spend more time with them.

> "I feel responsible for my children and grandchildren and possibly my great grandchildren having this condition because it started with me, it must have come from one of my parents but they're not here now."
>
> Survey respondent

> "I'm very fortunate I have amazing parents who go above and beyond to help me but I always feel I'm in the way and their life would be easier and better without me and my struggles."
>
> Survey respondent

Fertility

A small number of survey respondents shared their experiences of having a rare chromosome or gene disorder had made starting a family more complicated, with some requiring fertility treatment to become pregnant. Some respondents expressed feelings of guilt that their partner was having to undergo IVF while it was them who carried the condition. Others shared the grief of pregnancy loss made worse by interactions with uninformed healthcare professionals who did not know about their rare condition.

> "When accessing support for recurrent miscarriage, we were constantly educating health care professionals about what it meant - even the genetic counsellor we accessed through the NHS seemed to misunderstand the statistics."
>
> Survey respondent

> "Guilt. I feel so much guilt being the reason my wife and I can't fall pregnant, and yet she is the one undergoing all the intense treatment and it's impacting her body because of something I have."
>
> Survey respondent

> "For me I feel like my biggest challenge is staying pregnant. I have had 3 miscarriages and only 2 full term pregnancies and I feel like one of the fortunate ones because I know that there are so many women who have had so many more miscarriages and don't have any living children or if they do, they have had so many more miscarriages than me."
>
> Survey respondent

WILLOW

Case study

My name is Willow Foster-Thorpe and I was born in 1998. I have had seizures since birth and a blood test found that I had triple X syndrome which means I have three X chromosomes instead of two. I had a bad seizure when I was 12 months old, it delayed my development, and alongside my triple X syndrome, led to learning and speech difficulties. I had to go to speech therapy from age 2 until I was 9 years old, and my mum had to take me out of school to go for speech and language therapy every week.

I wasn't at the same level of development as the other children in my classes and so over the years I really struggled to read, write and learn new things in lessons. I was also a very nervous, anxious and extremely shy child and didn't have any confidence. I would get upset a lot and cry very easily. My mum had to fight to get funding for me for a teaching assistant to support me and she always had to go to lots of meetings.

When I was at school, I would never have imagined that when I grew up, I would become a writer and publish three books as I struggled to spell due to my dyslexia. Thanks to my mum for getting me into a specialist college. It was during my art classes there that I thought of creatures with different disabilities. Other students with different disabilities inspired me as, even though they had disabilities and bad health problems, they had courage and determination to get on with their lives and learn new things just like every other young person.

JESS AND TYRON

Case study

We recently reminisced on the moment we decided to start trying for a family, three years ago. It was exciting and we were so hopeful. We can't help to feel a bit naive to have ever thought getting pregnant would have come smoothly, but we never could have predicted the fertility rollercoaster ahead of us.

Ultrasounds confirmed my womb was healthy and ready to welcome a bubba, and a laparoscopy eradicated my endometriosis. Several months later and we had never seen double lines. We did routine GP tests, which led us to discover my husband, Tyron, had severely low sperm count. We were immediately referred to a fertility specialist, and further investigation uncovered he carried a balanced reciprocal translocation (BT), where his chromosomes 4 and 19 are crossed. My husband was healthy in every way, except he will have difficulty trying to conceive a child. This condition, present since birth, impacts his sperm count, morphology, and motility.

We dove straight into IVF, aiming for at least three to four viable embryos before attempting a transfer. After four cycles we achieved one beautiful, viable embryo! It gave us immense hope and determination. We finally had confirmation that it was possible to achieve an embryo that was half me and half Tyron.

Over 17 months we completed eight IVF retrievals, collecting 131 eggs that resulted in 11 blastocysts. We ended 2024 with four precious embryos – three euploid and one complex mosaic. The euploids carry Tyron's BT, meaning they will face reproductive challenges in the future.

Our journey is not over, but we are hopeful for what 2025 will be for us. We try our first transfer soon and are immensely grateful to have reached this stage.

KEY FINDING 3

Individuals and families feel isolated

Having, or caring for someone with a rare chromosome or gene disorder can strengthen close family relationships and bonds. However, managing grief, living with constant uncertainty, the complexities of care and managing the condition often negatively impact on social networks and relationships resulting in significant feelings of isolation and loneliness.

Many of the parents/carers who completed the survey or participated in the focus groups were providing high levels of care for their children. This often included managing complex medical and/or behavioural needs, juggling appointments and navigating health, education and social care systems. They were also managing the emotional impact of grief, uncertainty, worry, stress and anxiety.

Affected individuals who responded to the survey reported how they often felt judged, misunderstood and managing their symptoms could be time consuming. They however reported higher levels of maintaining social networks than the parent/carers who responded to the survey.

Respondents and survey participants shared how their immediate family relationships were very close, but lack of awareness, managing the practical demands of care alongside the emotional demands of coping with uncertainty and grief often resulted in feelings of isolation. Either it was too much trying to manage everything required for social activities, and/or it felt too much overcoming feelings of anxiety, dread and fear of judgement.

69% of affected individuals felt they had maintained good social networks and spent time with friends.

34% of parents and carers felt they had maintained good social networks and spent time with friends.

"Having a child/young person with higher needs than their siblings, both physically and especially behaviour wise. It causes extreme difficulties when planning family events/ outings."

Survey respondent

Isolation

998 respondents (61%) agreed with the statement 'I feel isolated' (23% strongly agreed, 38% agreed). From the survey responses mothers/stepmothers of affected children reported feeling higher levels of isolation than fathers/stepfathers.

Isolated was also one of the top five words selected by survey respondents to describe their experiences. Other related words included:

- Lonely
- Misunderstood
- Stigmatised

For parents/carers of adults, the isolation they experience intensifies their fears for their child's future once they are no longer able to care for them.

"And I think the overwhelming feeling when your daughter is one of 19 with a condition is really intense isolation. I think you think you're the only family in the world."

Focus group participant

"I've always just looked after my daughter on my own. But I completely feel isolated and worried for the future. That's all I feel every single day. It's just always on my mind about what's going to happen in the future to her."

Focus group participant

59% of affected adults agreed they felt isolated.

47% of fathers/stepfathers agreed they felt isolated.

68% of mothers/stepmothers agreed they felt isolated.

Uncertainty and anxiety

Throughout all the survey responses and in the group discussions the anxiety caused by uncertainty about the condition and worries about the future was a common theme. For parents of young children, this uncertainty often related to trying to make sense of the diagnosis and what this meant for their child's development and future.

Parents of adults, however, were concerned about the uncertainty of what will happen when they can no longer care or advocate for their child. They also worried about possible declines in their child's health as they aged as there was little information available about what to expect.

Anxious was the top word chosen by survey respondents to describe their experiences.

Related words chosen included:

- Sad
- Depressed
- Angry

"I feel the biggest challenge for family's affected but rare chromosomes and gene disorders is the uncertainty. Uncertainty of if they'll ever be able to do what everyone their own age can do. If they'll cope in school. If they will make good friends. If they will be able to work and live independently. Behind every corner is a big wave of uncertainty. But when they reach milestones even if they're behind it's pure and sheer excitement and a sense of pride that other parents wouldn't understand."

Survey respondent

"In our particular case, the consequences have not been that bad so the worst is the uncertainty that comes with the syndrome. Will he keep developing more or less at the same rythm as he has so far? Will there be a moment when he falls behind in terms of learning/cognition? Will other health or mental health issues come out in the future? Will he be able to have a 'healthy' child if he wishes to in the future?"

Survey respondent

"The older the young person gets the more frightening the future is."

Survey respondent

Grief

Most of the group discussions touched on grief and the different ways this can manifest. It was also identified by some survey respondents as one of the biggest challenges. For some it was grief for the loss of the parenting experience they expected, for some it was anticipatory grief of how their child's life may be, or if it would be shorter. Others grieved for the experiences their child would not have.

"The most difficult thing for me right now is the unpredictability for the future: is he going to cope with seizures? Will he ever be able to look after himself? Will we have to support him and be with him, the unpredictability and the different future that we wouldn't have expected, are all new considerations for us these days?"

Feedback session participant,

"I see people of her age doing stuff and I think ...R will never experience love like this. You know, R will never get married...will never have children .. over the years, you do grieve for different things that you imagined, or you'd wish for them ... but, you know, obviously R's understanding is so limited that, you know, there's a lot that R will never experience and that you know, that makes me sad."

Feedback session participant

"Constant grief for what could have been, what is and what will never be. All at different times."

Survey respondent

Family relationships

Overall most survey respondents and group participants reported close family relationships, with good emotional and mental wellbeing and that they enjoyed quality time together. Many group participants talked about the impact on siblings and how their experiences and the impact on them can often be overlooked, but also how positive they could be.

Couple/parent relationships

Many of the survey respondents and group participants reported that their relationship was strengthened because of the shared experience of managing a rare chromosome or gene disorder in the family. Others shared how their relationship had broken down because of the pressures of managing the impact of diagnosis and/or caring for their child.

> "Selfishly, the hardest part of this hasn't been the countless surgeries and the many unknowns, it's been the loss of relationship with my partner. We process the diagnosis and treatments so differently, and with how rare our child's gene deletions are, even the family therapist felt she couldn't really understand the depths of our struggles. That was a wild thing to hear."
>
> Survey respondent

> "No one ever really can understand how difficult my life is apart from my wife."
>
> Survey respondent

Extended family relationships

Survey responses were balanced relating to how easy people found it to explain their, or their child's rare chromosome or gene disorder. In the focus groups many of the participants discussed how they struggled with extended family members who did not always understand the impact of the rare chromosome or gene disorder. For some this meant they had withdrawn from these wider family relationships and just focused on their more immediate ones.

> "Some of my family - my parents in particular - have been very supportive and understanding. Other members of the family see my son as naughty and uncontrollable and hold higher expectations for him."
>
> Survey respondent

> "We also had one set of grandparents and some relatives who did not include us in family gatherings."
>
> Survey respondent

> "No one can really understand if they haven't experienced it (we worked out we have over 100 hospital appointments a year, at least just for my 2 children with rare disorders). It causes a lot of stress within our extended family because we are asking for childcare so we can attend appointments."
>
> Survey respondent

ANNIE
Case study

Our family was blessed when we welcomed our daughter Annie into the world. She inherited a deletion in her CDK5RAP2 gene from her father and I, causing her to develop primary microcephaly. We had so many concerns when we found out about her diagnosis during my pregnancy. We would have been so very reassured if we knew then what we know now about our gorgeous girl.

Annie is a social butterfly, making friends wherever she goes. She shows lots of care, affection and empathy for all beings. She enjoys breakdancing, painting, water play and reading lots of books. We love spending the day at the beach or down the park. Annie attends daycare and is just about to begin kindergarten at her local school. She engages in speech therapy, occupational therapy and physio weekly to work on her skills and abilities. Her childcare centre employs an additional staff member to support her during the day, using funding from the government. We have applied for support at school as well. Annie experiences sensory overwhelm and finds changes to routine very disorienting and stressful at times. Shes very attached to her family, and becomes dysregulated if she doesn't get enough quality time or affection. We have lots of 'huggles' and playtime throughout the day. She's our rare precious gem and we love her dearly.

Social networks

46% of survey respondents felt they had not been able to maintain good social networks and spend time with friends.

Survey comments and discussions in the groups suggested there were a number of barriers experienced by families including lack of understanding of the impact of the condition, lack of suitable places to go and feeling alienated from others.

Many parents of adults commented how it had got harder as their children aged to find suitable activities or ways for them to be part of their community.

Of the 34% of survey respondents who felt they were able to maintain good social networks and spend time with friends, there seemed to also be a correlation with higher reported rates of individual emotional wellbeing (55% compared to 39% for all responses) and family wellbeing (64% compared to 40% for all responses).

It's been hard work to help eliminate or at least minimise the risk of other parents withdrawing from having our daughter visit due to 'fear' of the unknown represented by her 'disability'. Most kids don't see disabilities, they see another potential friend, but adults have a tendency to look for obstacles and fear them."

Facebook response

Contact with other rare families

Although most group participants shared how valuable they found it to be in contact with other families affected by rare chromosome and gene disorders, 56% of survey respondents had found it difficult to connect with other affected families/people.

For me the main thing in the early days, and still now, is that I'm desperate for like a local connection because I've got my mum friends from my other two kids and I've got this brilliant community and they're all amazing, and they're really supportive. They're brilliant with like understanding as it can be, but they don't get it because they haven't, they're not living it, so I just desperately want some connections with parents locally, so I can just, you know, go for coffees and the kids can play together as well because I'm really mindful that I mean, I don't know where she'll end up developmentally, but it'd be nicer to have people to grow up with. You know like, they aren't all going to move or go to uni and leave her, do you know what I mean?"

Focus group participant

Case study

REYNOLDS FAMILY

I won't forget the day we received our daughter's diagnosis of 22q11.2 deletion syndrome. It was a whirlwind of emotions – shock, disbelief, and fear for her future. This rare chromosomal disorder affects approximately one in 4,000 births, and its impact can vary widely from person to person.

For our family, it meant facing a myriad of challenges. Our daughter spent much of her first year in the hospital, undergoing two open-heart surgeries. The road to recovery has been long and arduous, filled with countless doctor appointments, therapies, and hospital stays.

Yet, through it all, we've found strength in community. Connecting with other families facing similar challenges has been invaluable. We've learned from each other, shared experiences, and offered support. These connections have become lifelines, reminding us we're not alone in this journey.

Our daughter is a fighter, a miracle. She has shown incredible resilience and determination. We celebrate her every milestone, no matter how small. We advocate for her needs and work tirelessly to ensure she receives the best possible care. Our family has grown stronger through this experience, and we are grateful for the opportunity to raise awareness and support others affected by rare chromosome disorders.

OLIVIA

Case study

"SOCIETY DOESN'T CARE ABOUT AGE IF THEY PHYSICALLY LOOK 'LITTLE'"

Developmental delay. That's what they called it until she was 6, then that label stopped applying for school, but not to the world. Society doesn't care about age if they physically look "little" but when that kid becomes four feet tall.... We essentially had a 3-year-old at home for 8 years, but no one noticed. Finally, Olivia's mental development began to grow beyond 3, but unfortunately so did her body. Now we have a socially awkward 7-year-old in a pre-teen body. People look up when the volume/tone doesn't match the sleek slender teen. They cock their head watching a teen drop to all fours acting as a cat. Before there were conspiratorial glances of understanding from parents about the tantrums. Others thinking they know what it's like to live with the terrible twos... But now it's anger because society is disrupted by a spoiled teen. It's not all bad though, as her milestones mean more to me. She began ice skating at 3; only marching on the ice for 3 years. Then learned to mimic, became graceful and amazing - but hit technical walls. Six months ago after another mental jump, she learned how to learn. "Mom, crossovers are hard!" she said, and I cried knowing her brain understood the truth.

KEY FINDING 4

Advocacy is not always valued or a choice

Many individuals and families become experts in the condition affecting them or their loved one, but their advocacy, insights and lived experience are not always recognised or valued by professionals. This can create additional pressures and responsibilities and can hinder collaborative care.

Experts by experience

75% of survey respondents felt they had become an expert by experience in the rare chromosome or gene disorder impacting them and/or their family.

67% of survey respondents agreed that they had felt able to advocate for themselves and/or their child.

However comments and discussions suggested this was not always a choice, there was simply no one else to do it.

With so little information available, and so little awareness or understanding from professionals, most people felt they had little choice but to find out as much as possible about their condition. In the majority of cases many were also coordinating care and ensuring all professionals were kept up to date and information was shared appropriately.

Throughout the survey responses and discussions, some positive themes emerged. Some survey respondents chose words such as determined, hopeful, focused, motivated, passionate and strong to describe their experiences. Other themes emerged relating to how their experiences had made them stronger – both as an individual and as a family, learning 'what really matters' and feeling enriched.

> "There's not much to know about it, its very rare and there's not many in the world who have it. My son has lots of things wrong with him so he has to see lots of doctors and health professionals and they've not heard of it so I have to explain it over and over again. We need more help and support with it. I took him to see a genectic doctor two weeks ago she was very good but you just get told about it and then your left to get on with it without any further help at all or support."
>
> Survey respondent

> "I feel like I have become the expert in my child's rare genetic disorder. Her medical team trusts me to convey information and to help make informed decisions (to the best of our ability) based on what I can share with them. This is both a blessing, but also incredibly overwhelming! I'm just a mom - I don't have an advanced degree in medicine or genetics, nor do I know how to read any of the articles that come out that even mention my child's deletion."
>
> Survey respondent

> "It is a tough journey but it has taught me some valuable life lessons. I'm more enriched as a person because of our rare chromosome family member."
>
> Survey respondent

Pressures of being an advocate

Many parent carers shared that advocating for their child had taken a toll on their own physical or mental wellbeing and that they often doubted themselves and worried that they weren't doing enough. Many were proud of what they had achieved for their child, but also sometimes felt overwhelmed by the responsibility.

Many expressed frustration at the lack of signposting from professionals involved in their child's care and that they were always left to research themselves. They also expressed concern that some parents may have their needs such as learning disabilities or language barriers which meant they would not be able to effectively advocate for themselves or their child. It was felt that in the absence of systematic signposting and pathways this again meant families were often reliant on 'luck' to access support.

> **"We advocate for our son because there's no one else."**
>
> Survey respondent

> **It's a really exhausting process and depending on their age, their developmental stage, their condition, you are literally putting your heart and soul into fighting. It's not just about being an expert, it physically takes it out of you as well."**
>
> Feedback group participant

> **So we advocate because there's no one else .. I get told something and then the next person comes along and they contradict the person that comes forward. Say Oh no, no, no. They're wrong. Yeah. So I'm having to be the person that knows so that I can turn around and say, no, you're wrong actually. And it's incredibly tiring. I mean, it's really, really tiring."**
>
> Feedback group participant

> **You don't know, and that's the scary thing, you know. You read and you read and you read and you read. But you're always wondering, is there something else I should know, or should have read?"**
>
> Feedback group participant

Lived experience/advocates not always valued

During the feedback sessions exploring the key findings there were a number of discussions about how parents felt that they were not always respected as their child's advocate. Some felt they had been forced to take on this role because of systemic failings but then were not valued by the professionals working with their child.

They shared stories of how they had been belittled or ignored at multidisciplinary meetings, and how their views were rejected because their child presented differently at school. Many felt that the language often used by professionals was inherently dismissive and undermining.

The consequences of this was the sense that everything was a 'battle' and a prime cause of the stress and frustration described earlier. Many noted that this had significantly impacted on their general sense of being and contributed to their isolation and alienation from their friends.

> **I mean, I know I've lost friendships kind of because people can't cope .. and then in terms of the fact that you are always battling for services, you do kind of very often, I know in my experience get a label for being someone who battles and all of that isolates you as well."**
>
> Feedback session participant

> **I think it's the way with chromosome abnormalities, you don't necessarily fit into a textbook, but you know your child better than anyone, and I agree with the saying, 'Mum knows best' but I don't think professionals always see it that way."**
>
> Feedback group participant

Adults, independence and mental capacity

Many parents of adults shared how it was at times challenging navigating relationships with professionals involved in their adult child's care. Most were still actively involved in providing, or managing, their child's care, but were not always respected for doing so.

They described how, as their child moved through transition and into adulthood, they felt they were often treated with mistrust and as a barrier to their child's independence. This was particularly challenging for parents of adults who were deemed to have mental capacity to make their own decisions, but lacked suitable support from social care/their independent living providers to make appropriate and safe ones.

> **The frustrations of dealing with social services when a vulnerable adult is deemed to have capacity 'with support' but who then refuses support and then anyone interacting with or trying to support her has to stand by and watch her make terrible decisions because according to Social Services she has a right to make mistakes. That only works so far when families are then left unpicking the mistakes or trying to correct what's gone wrong."**
>
> Survey response

Summary and our commitments

Over the next five years Unique commits to investing in our team to increase our outreach capacity and develop a strategy to increase the visibility of rare chromosome and gene disorders at a local, national and international level.

Unique is a UK based charity but due to the rarity of the conditions we support, our registry and community is by necessity, international. While some challenges faced by the individuals and families we support may be specific to the national context and healthcare system, there are universal experiences such as living with uncertainty about a condition, coping with judgement and lack of understanding from others, and scarce information that cut across borders.

Throughout all the stories we heard was a common theme, the issues individuals and families faced were often caused, and/or exacerbated by, the lack of awareness of their rare chromosome or gene disorder. Greater public and professional understanding would go a long way to reducing some of the key issues faced by families and individuals affected by rare chromosome and gene disorders, and in doing so, perhaps creating space instead for acknowledging and celebrating their achievements despite the challenges they face.

Over recent years our focus at Unique has been primarily on providing much needed information about rare chromosome and gene disorders to fill the information void. We now have over 300 condition specific guides and throughout our consultation participants confirmed how these had been a life-line in the early days. We will continue to update and expand these guides, but we will also now look more broadly at how we can improve the lives of our community by striving for greater visibility of rare chromosome and gene disorders.

We will:

Develop new information and advocacy resources to empower our members to educate and raise awareness of rare chromosome and gene disorders in their local areas or countries.

Work with our partners in the genomics community to embed understanding of lived experience of rare chromosome and gene disorders in research and clinical settings.

Engage with policy makers to ensure rare chromosome and gene disorders are equally represented alongside more well known conditions such as Down's syndrome and autism spectrum disorders.

In the UK we will run a targeted education and outreach programme to increase awareness, diversity, inclusion, understanding and representation of rare chromosome and gene disorders within Unique membership and throughout health, education and social care. We hope the findings can be employed and rolled out around the world.

Thank you to everyone who took part in our survey, attended our focus groups and feedback sessions and to those who engaged on social media and submitted case studies for the report and Little Red Book.

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UNDERSTANDING GENES & CHROMOSOMES