Unique: The Rare Chromosome and Gene Disorder Support Group

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About me

- I have worked for Unique for 4 years
- I offer emotional support and practical information to families and professionals
- I have recently finished my MSc in Genetic and Genomic Counselling from Cardiff University
- Previously I have worked in research including a genetics and mental health study called IMAGINE ID, and research in improving access to palliative and end-of-life care

Presentation Outline

- Who we are at Unique
- About our members
- Unique's work with families
- The emotional and social impact of a diagnosis on the family
- Unique's information resources
- Unique's work with professionals
- Awareness Day

Who are *Unique*?

- *Unique* is a registered charity who rely solely on public donations
- Made up of a team of 8 highly-motivated staff and board of Trustees
- Established in 1984 by Edna Knight, *Unique*'s Life President and one of our Trustees
- Registered over 27,000 families from 118 countries worldwide on *Unique* Database Registry (40% in the UK)
- 150-200 new families join every month

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Unique's Mission

- **"Together, we will beat the isolation of rare chromosome and rare gene disorders"**

- As one of many support groups and charities representing the **rare disease community**, Unique's mission is to **inform, support and alleviate the isolation** of anyone affected by a rare chromosome or single gene disorder and to **raise public awareness** of these conditions.

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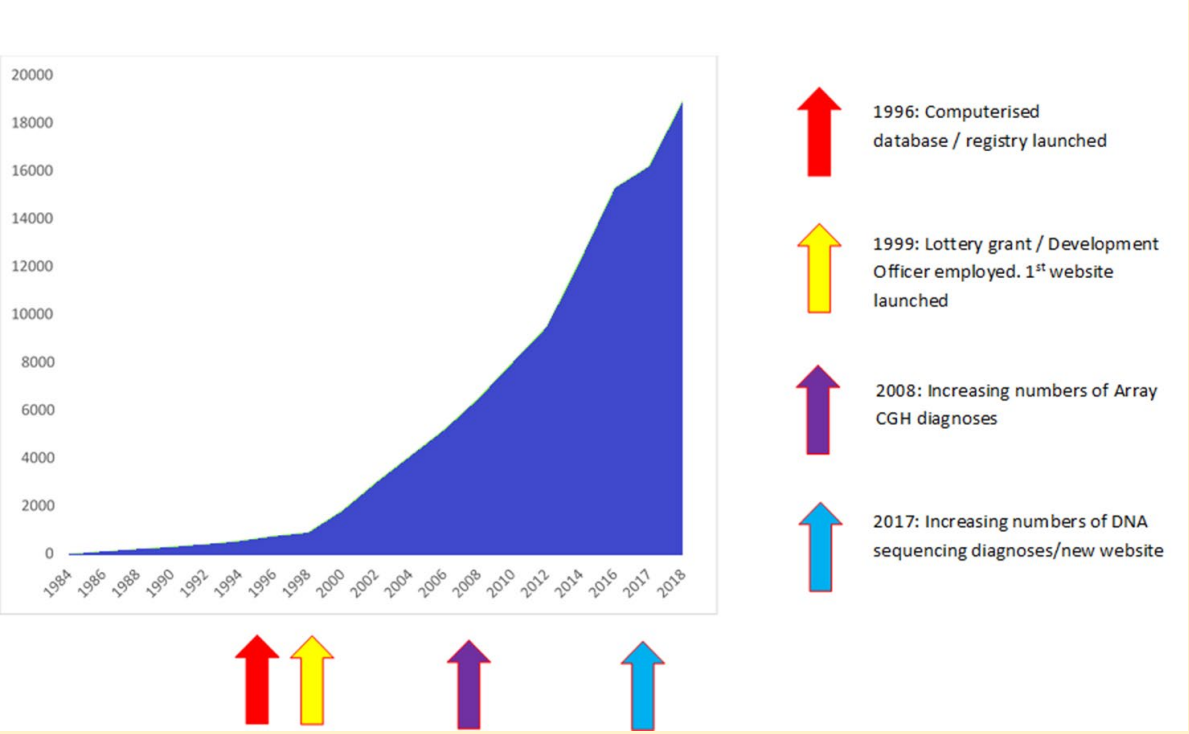
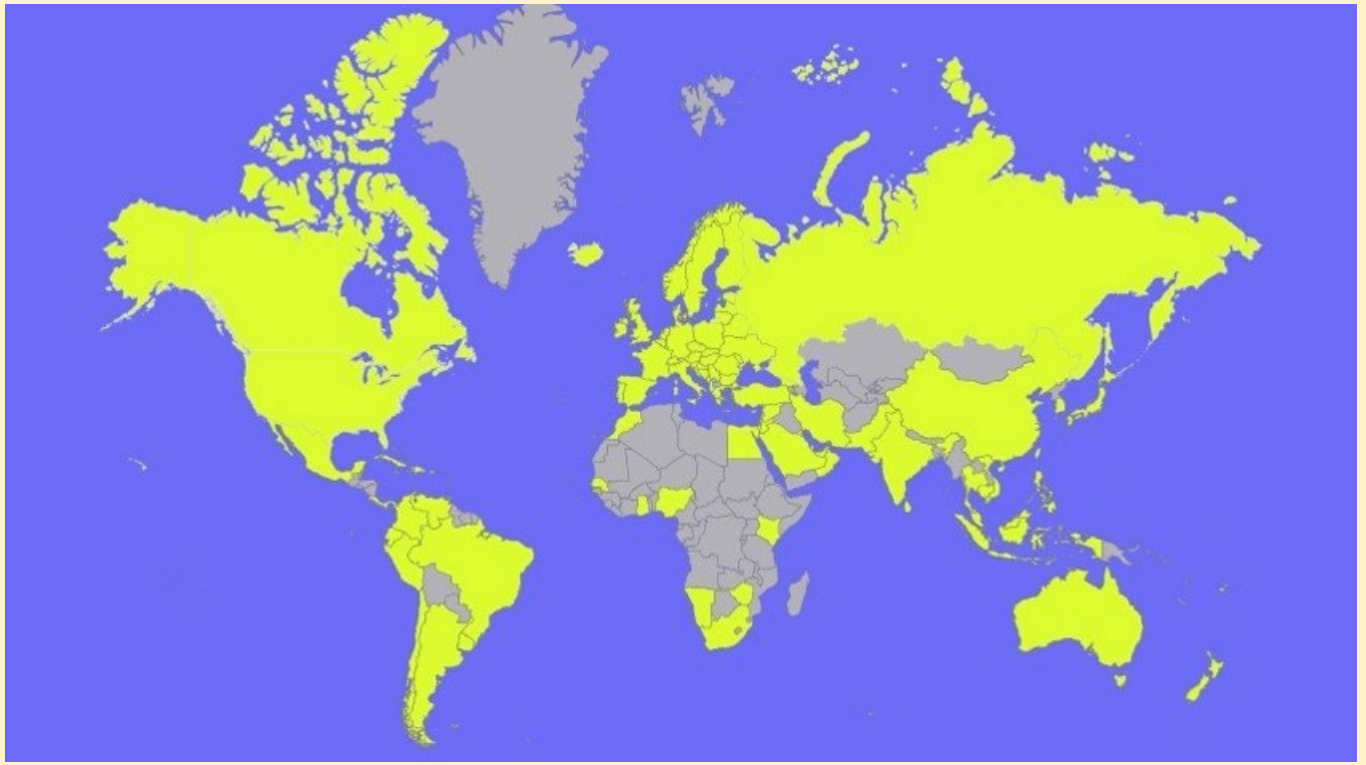
Unique's Membership Growth



- The increasing number of families receiving a diagnosis as a result of **advances in genetic testing** has had a massive impact on our membership numbers.

Unique's Global Membership



At November 2022, 27,200+ member families representing 29,400+ individuals diagnosed with a rare chromosome or gene disorder in >110 countries; 40% in the UK.
150-200 new families now joining each month.

Unique's Membership of DYRK1A families

Country	Number of members
UK	16
USA	5
Australia	2
Argentina	1
Republic of Ireland	1
Poland	1
South Africa	1
Switzerland	1
Total	**28**

Please do join as a member if you haven't already, we would love to welcome you to the Unique community!

What Unique can offer families

Informed & helpful signposting

Information guides
specific to genetic diagnosis or mechanism of disorder, signposting

General & Practical guides
covering day-to-day living and genetic testing techniques

Free membership
onto the *Unique* registry database

Regular magazines & updates
covering new guides, fundraising events and research

Emotional support

Contact details
consenting member families & organisations

Private chats
with *Unique* staff on our helpline & social media groups

Family days & events
& online Q&As with experts

A sense of community & belonging
through good and bad times

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What makes all our members Unique?

A chromosome or gene disorder can impact health and development in complex ways

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Why a diagnosis is important to families

Curtailment of the ‘diagnostic odyssey’

(multiple referrals/investigations)

Explanation and name for their child’s symptoms

Access to better information and support

Research stimulus and data sharing

Finding others with a similar disorder and experience

Peace of mind – their concerns are real

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While a diagnosis can come as a relief, it can also lead to a mix of emotions…

"How did it feel when you received the results of genetic testing?

Overwhelming relief – I'd always blamed myself for something that I must have done wrong during pregnancy or birth. I was upset and tearful for about a week or so. I was shocked. Then I was amazed to find other children who looked like my son when I'd spent years looking for them!

Almost relieved at first to know that there was an issue, also shocked and upset. Then hopeful that this may lead to help for X in the long term and to understand him much better

While a speedy and accurate diagnosis can bring great solace, comfort, information and support to families, formal Genetic Counselling of the highest quality is essential before testing and after diagnosis.

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The impact of a diagnosis – how professionals communicate REALLY matters!

How results are explained to families has a big impact and comes with its own challenges

- Lack of recognition of the disorder by other professionals

"They won't believe me"

"What does this mean for my child?"

- Lack of evidence and accessible information

"Why is there such variability in how people with the same diagnosis are affected"

- Variable expressivity

"Why can't they be sure? Its such a worry"

- Variants of uncertain significance

The psychological and social impact on siblings?

- Research conducted as part of my MSc dissertation
- Through Unique membership adult siblings of individuals with rare genetic conditions associated with a developmental disorder were invited to take part
- 18 adult siblings (aged 18-70y) from across the world consented and were interviewed
- Interview data was analysed using thematic analysis

Results of Thematic Analysis: (confidential unpublished data)

- Overarching theme of **Acceptance**
- **Altruism**; leading to compensatory, appeasing and protective behaviours. Often embarked on 'helping careers'
- **Normalisation**; this was their reality and normal, they felt proud and united, but acknowledged there was an established hierarchy of needs in the family where their affected sibling came first
- **Self-discovery**; adults tentatively separated from the family when growing up, this led to some feelings of guilt and grief
- **Devotion**; siblings were used as relationship barometers to vet new connections, they were expertly attuned to their siblings' needs and used these skills to translate and advocate for them
- **Anticipation**; uncertainty of the future could lead to feelings of anxiety which adults tried to cope with by controlling the situation or environment as best they could. Some uncertainty surrounded their siblings' diagnoses and inheritance pattern, including the chances of them having an affected child even when the inheritance pattern of their siblings' diagnosis was known to be *de novo.*
- **Frustration** (only theme not connected to Acceptance); stemmed from societal ignorance and healthcare abandonment

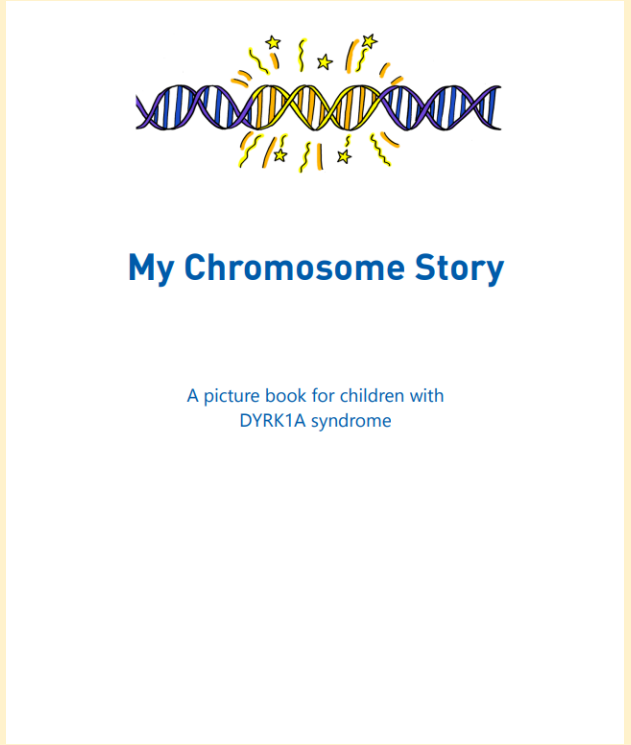
Next steps

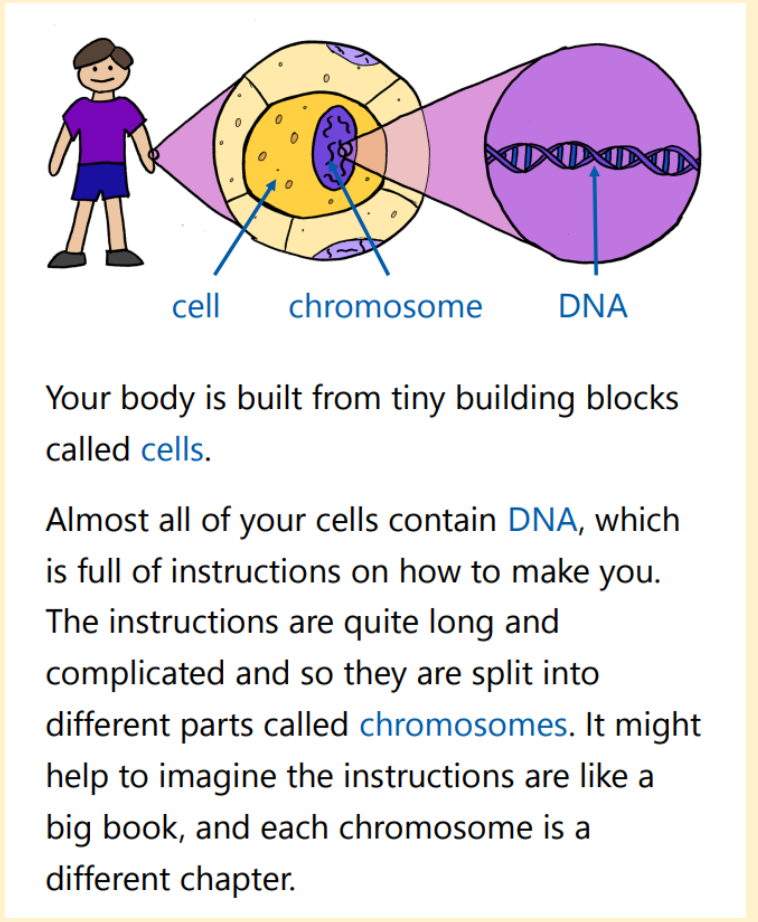
- Presenting findings to the Annual Genetic Nurses and Counsellors conference in September 2023
- Publish the findings in an academic journal – e.g. Journal of Genetic Counseling
- Recommendations for sibling literature and resources to be made available in clinical genetics departments
- Clinical recommendations of supporting, involving and caring for siblings when a rare genetic diagnosis in the family is made (regardless of inheritance pattern)

Unique Information Guides:

- DYRK1A and 21q22.13 deletion syndrome guide – updated this year
- Available to download for free from Unique's website
- Also making easy-read formats and story booklets

We would love to know your opinions about these new booklets!





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How do families and professionals make use of our guides?

- **Families**
- Validation of concerns
- Validation of the existence of their child's diagnosis
- "Not alone"
- Detail shared experiences & symptoms
- Useful tool to help explain diagnosis to others
- Evidence at tribunals/panels
- Guide to what might happen & what to look out for
- Practical suggestions and tips
- Acknowledgment of the uncertainty and variability

- **Professionals**
- Used by lab-based geneticists, clinical geneticists, genetic counsellors, non-geneticist clinicians and other involved professionals worldwide to help counsel their patients/clients and to inform their own practice

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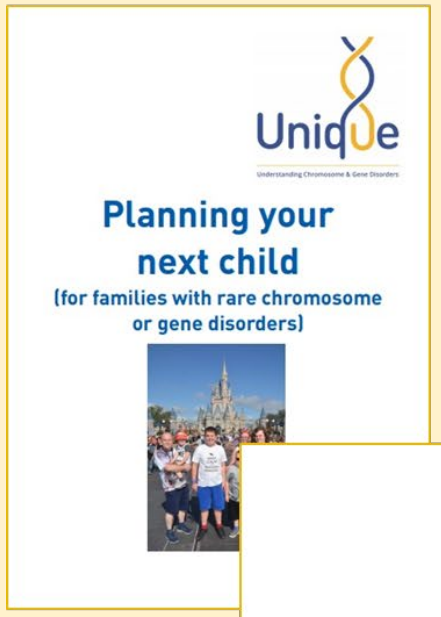
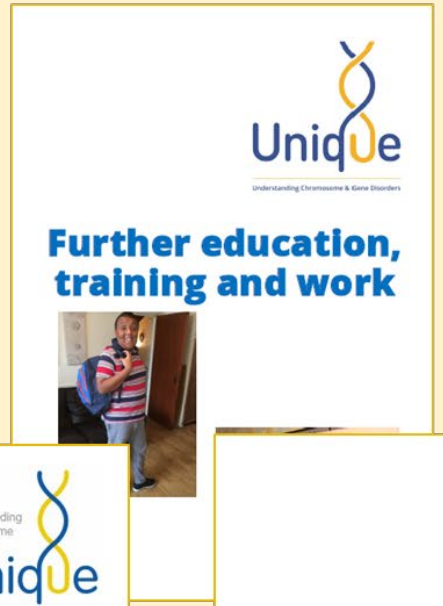
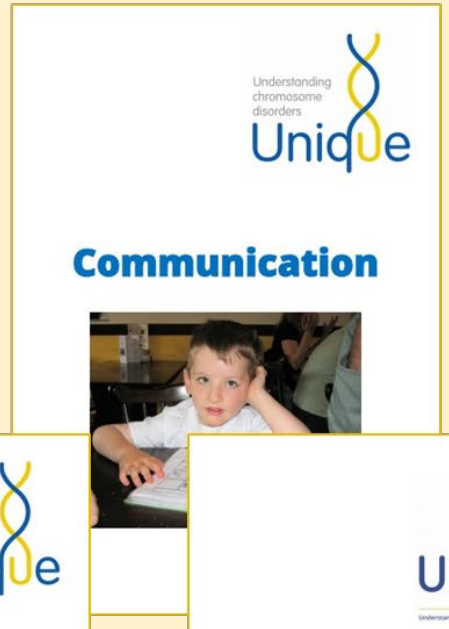
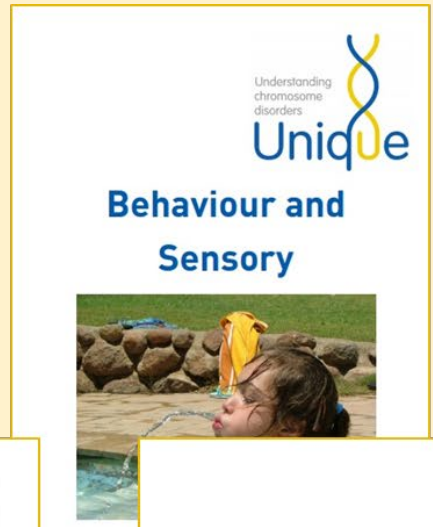
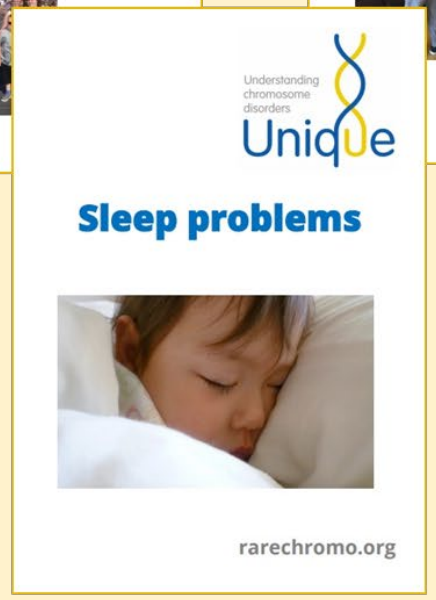
Unique Practical Support Guides - useful for all families!

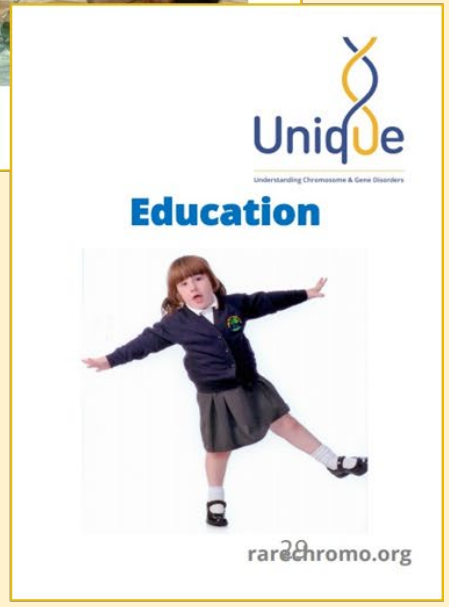












Unique working with Professionals

- Participating in national & international research
- Participating in professional initiatives and on advisory boards e.g. NHS England 100,000 Genomes Project Programme Boards, Rare Disease Transition Working Group
- Presenting the views of families to decision-makers
- Working & campaigning with other support organisations e.g. GAUK, SWAN
- Awareness-raising presentations/teaching sessions/study weekends
- Producing information guides e.g. reviewing, translations

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Awareness Day

- A day for us all to celebrate and educate, shine bright together and reach out to those who might not have heard of chromosome and gene disorders and need support
- We celebrate all the unique children and adults and their amazing achievements
- Good opportunity for members to share their experience with their families and wider community
- Coincided with the one-hour special BBC TV show ‘There She Goes’
- Next one in June 2024

Resources:

- **Our Unique webite**: https://rarechromo.org/
- **Disorder guides**: https://rarechromo.org/disorder-guides/
- **Family guides**: https://rarechromo.org/practical-guides-for-families/
- We are on **Facebook**: https://www.facebook.com/Uniquerarechromosomedisordersupportgroup/
- Instagram: https://www.instagram.com/unique_charity/
- YouTube: https://www.youtube.com/user/Rarechromo
- Twitter: https://twitter.com/unique_charity
- TikTok: https://www.tiktok.com/@uniquerarechromo

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Thank you for listening. Any Questions?

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