Unique:

The Patient Perspective

Sarah Wynn, CEO

www.rarechromo.org

Who are Unique?

- *Unique* is a registered charity who rely solely on public donations
- Established in 1984 by Edna Knight, *Unique*'s Life President and one of our Trustees
- Registered over 28,000 families from 120 countries worldwide on *Unique* Database Registry (40% in the UK)
- 150-200 new families join every month

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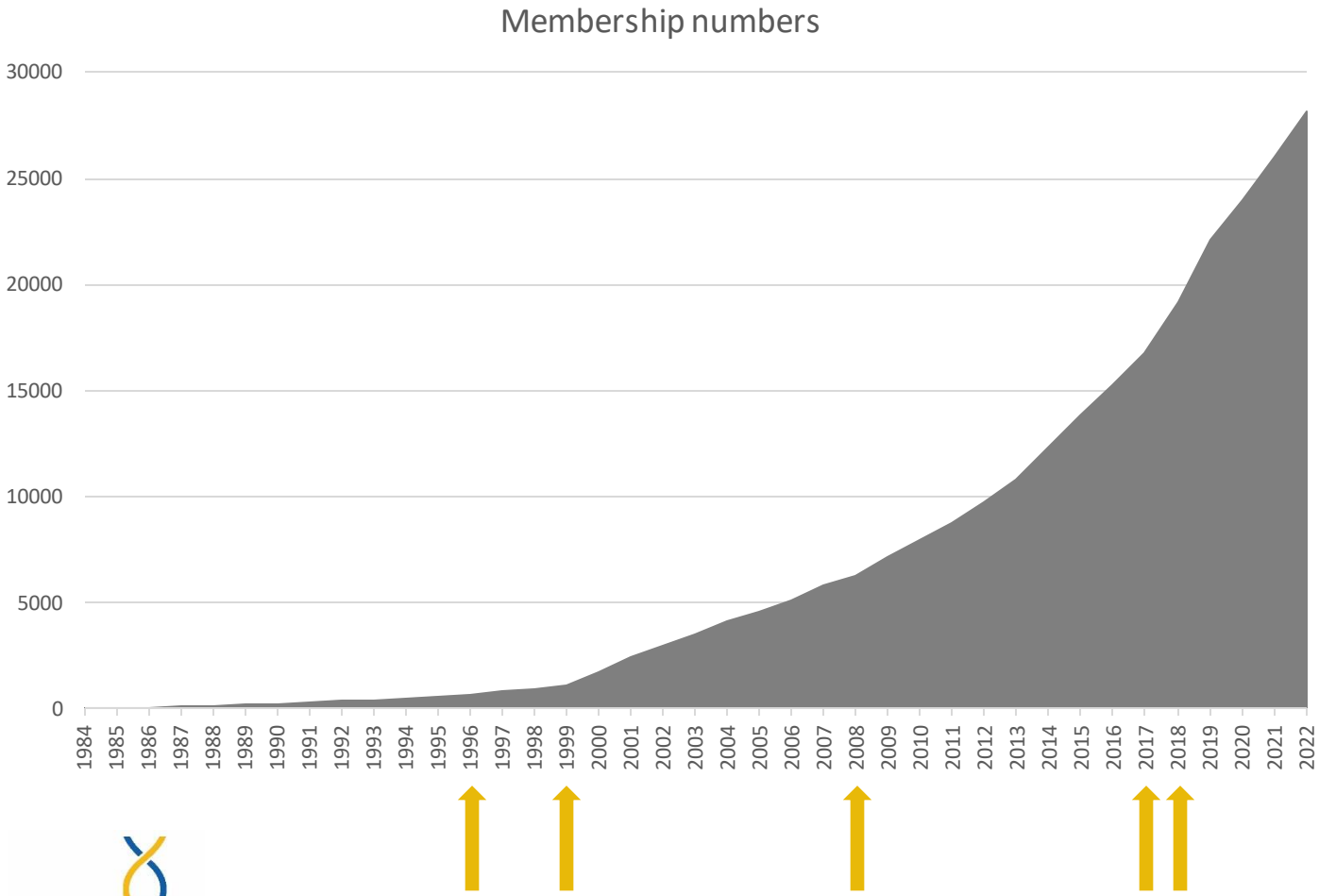
Unique's Membership Growth



- 1996: Database launched
- 1999: 1st employee & website launched
- 2008: Microarrays become common 1st line test
- 2017: New website launched
- 2018: NHS Genome Medicine Service (GMS) launched

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Unique's Mission

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

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Rare Conditions

- In the UK, 1 in 17 people will be affected by a rare condition during their lifetime.
- More than a third of people with a rare condition having to wait more than five years for a diagnosis

What patients and families want:

- Fast and accurate diagnosis.
- But also support along that journey

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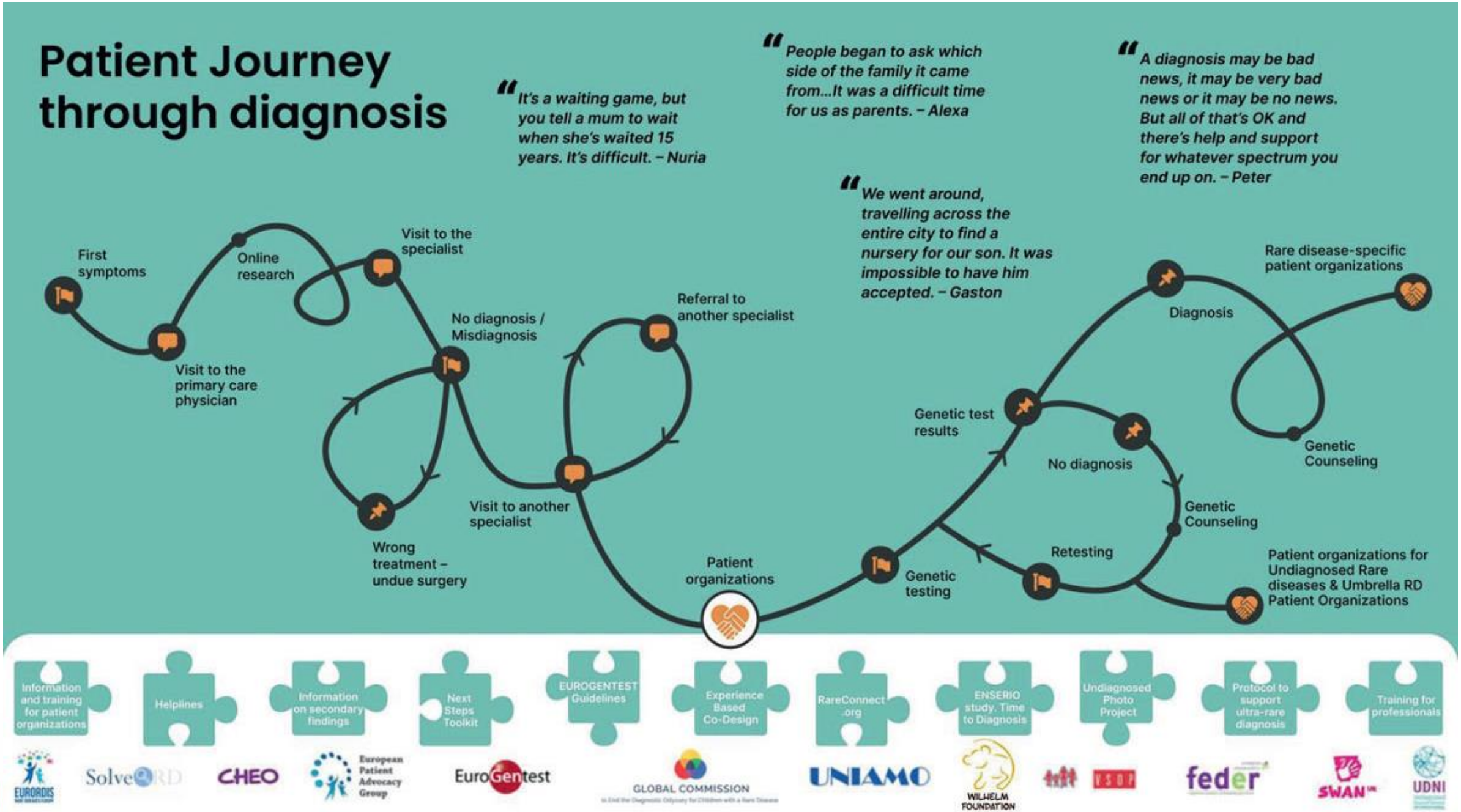

Infographic by Solve-RD Community Engagement Task Force, led by EURORDIS solve-rd.eu/community-engagement-task-force/

Why is a diagnosis important to families?

Curtailment of the 'diagnostic odyssey'

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

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The impact of a diagnosis

How do families even begin to describe their child's diagnosis to other people?

XXXY – it looks like a bad hand at Scrabble!

How do I explain my baby's microarray result to other people?

What's it called?

What on earth does this mean for my baby?

arr[hg19] 5p12q11.2(42719825-50991113)x3 13q11q12.12(1943628623373570)x3
17p11.2q11.2(20095030-25943693)x2~3
20q13.2q13.32(51873117-57136140)x3
arr[GRCh37] 16p11.2(28825605_29043450) x1 dn ID1B c.4148delCp.(Pro1383fs)dn

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There She Goes

- BBC comedy drama written by Shaun Pye and Sarah Crawford
- https://www.bbc.co.uk/iplayer/episode/m001n4rf

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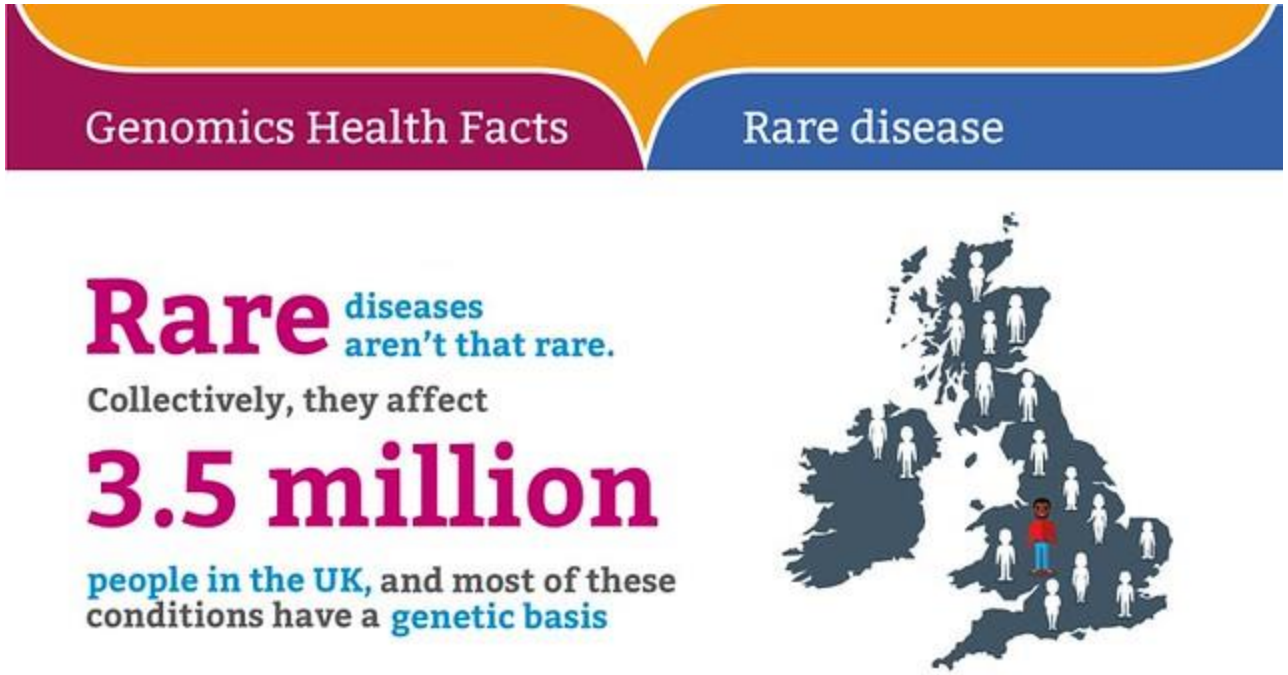
The impact of a diagnosis: rarity

- Lack of prognostic information
- Lack of a pathway to follow
- Struggle for recognition
- Challenges finding support
- Ignorance of professionals
- Lack of evidence-based treatments
- Lack of research



Health Education England's Genomics Education Programme (GEP)

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The impact of a diagnosis: uncertainty

- Rarity leads to a lack of certainty about what the diagnosis means for their child
- Variants of uncertain significance (VUS)
- Families feel isolated – not knowing where to turn for support and information
- Families feel helpless – how can they support their child

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What Unique can offer families

Information guides
specific to genetic diagnosis

Practical guides
covering day-to-day living and genetic ing techniques

Information & signposting

Free membership
onto the Unique registry database

Regular magazines & updates

Contact details
member families & organisations

Helpline
Telephone and email

Emotional support

Family days & events
local conferences & Q&As with experts

Community & belonging
through good and bad times

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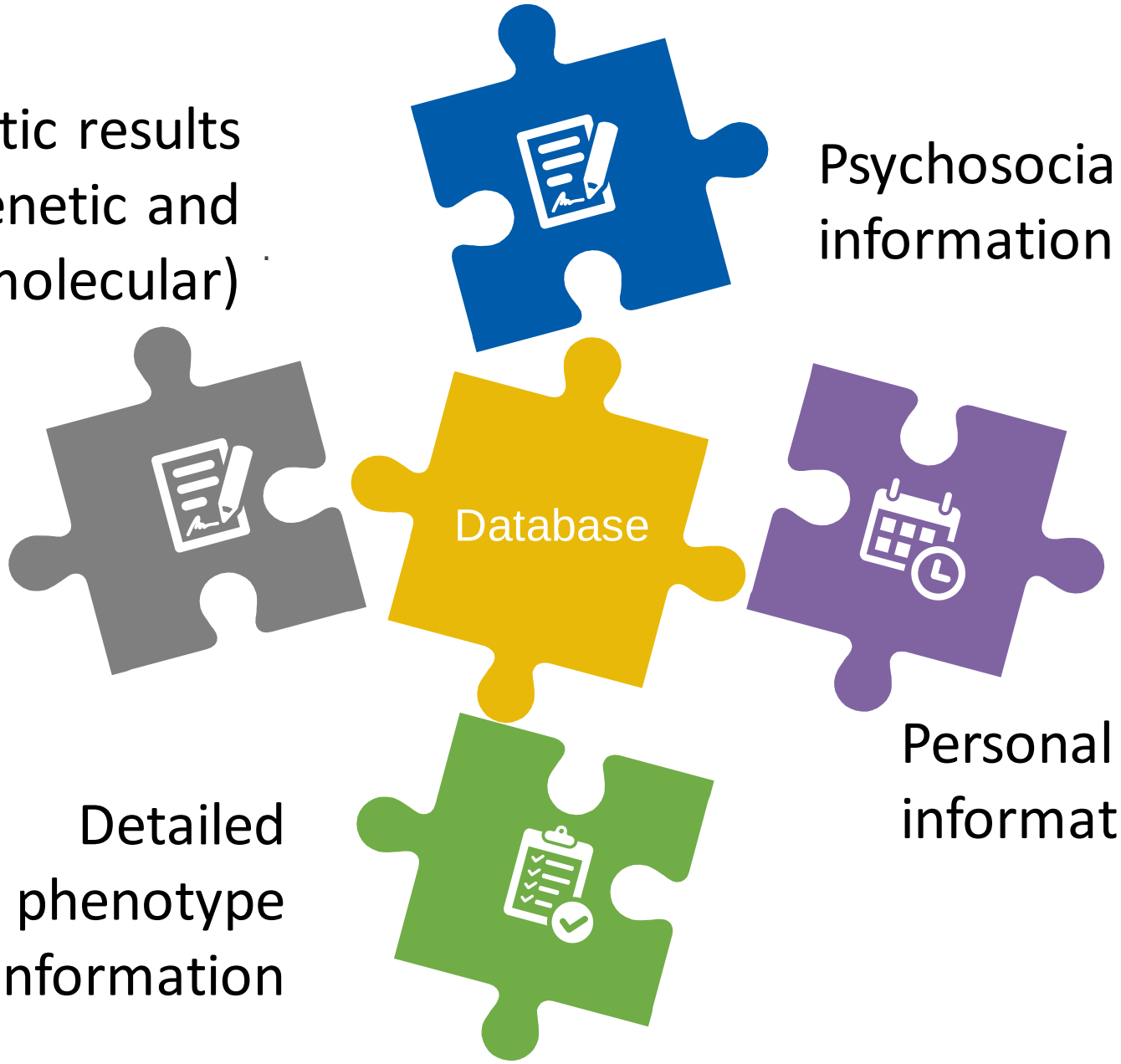
Unique database

Genetic results (cytogenetic and molecular)

Psychosocial information



Personal information

Detailed phenotype information

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Unique disorder guides for families and professionals

- Over 300 guides to genetic conditions
- Developed in collaboration with clinical and academic experts
- Translated into 22 languages

https://rarechromo.org/disorder-guides/

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Single Gene Conditions

- In collaboration with expert clinical geneticists
- Free to download from Unique's website
- Over 70 leaflets published
- More in development!

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www.rarechromo.org

How do families make use of our guides?

- Guide to what might happen & what to look out for
- Practical suggestions and tips
- “Not alone”, recognition of shared experiences & symptoms
- Validation of their concerns
- Validation of the existence of their child’s disorder
- Useful tool
- Evidence at tribunals/panels

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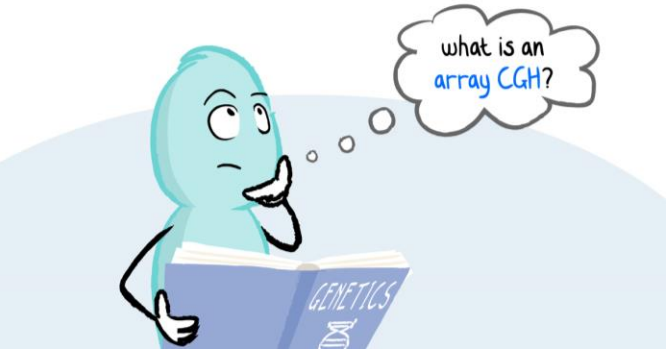
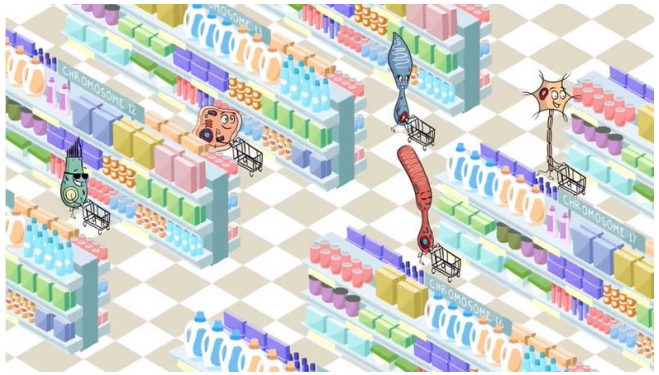
Other information

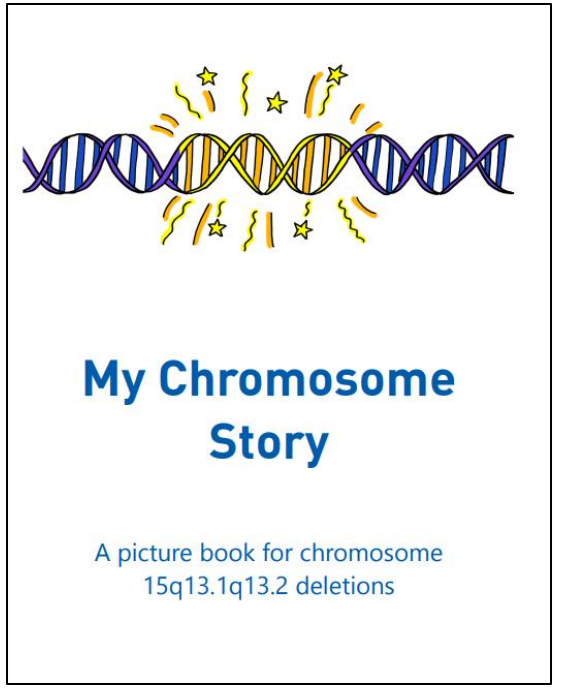
Animations:

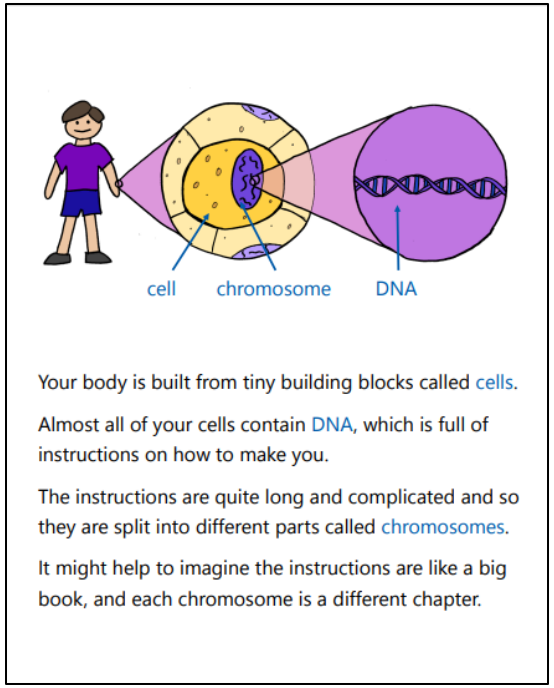
- DNA sequencing and ArrayCGH

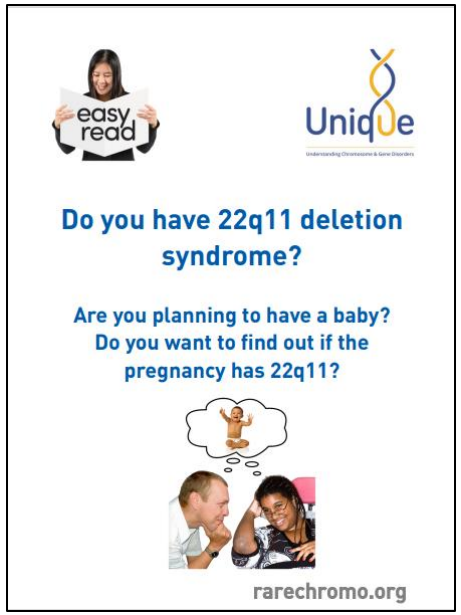




My Chromosome Story







EasyRead Guides

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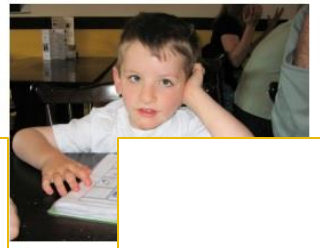
Practical Support Guides
- useful with & without diagnosis!

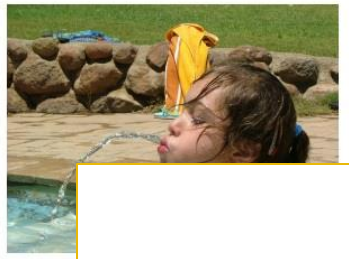
Planning your next child
(for families with rare chromosome or gene disorders)

Further education, training and work

Communication



Behaviour and Sensory



Sleep problems

rarechromo.org

Feeding guide for children with rare chromosome disorders

rarechromo.org

Carers Wellbeing

rarechromo.org

Education

rarechromo.org

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Thank you for listening!
Any questions?

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