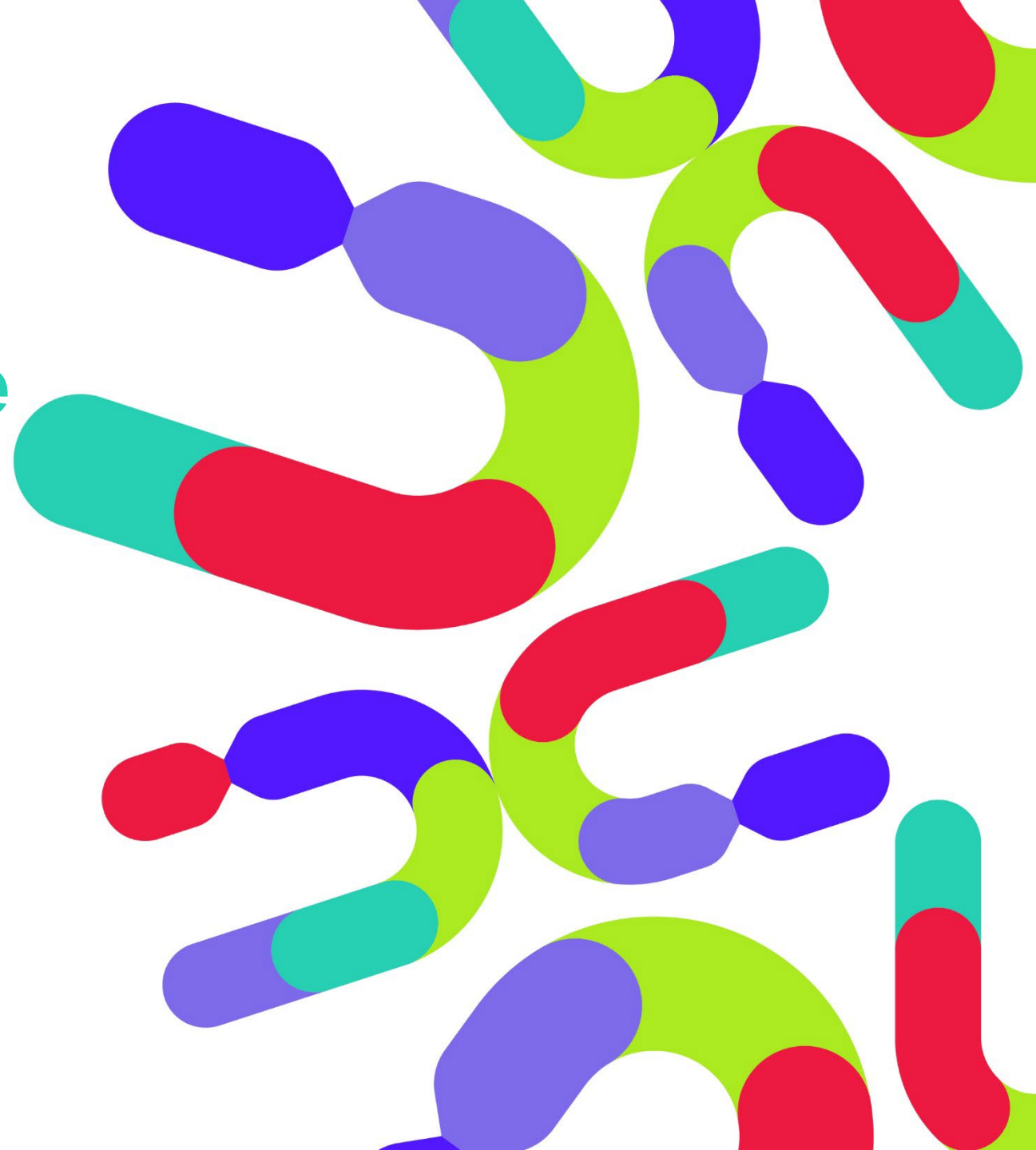


Unique: Rare Chromosome and Gene Disorder Support Group



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.




Who are Unique?

- A registered charity that relies solely on public donations
- Established in 1984 by Edna Knight, Unique's Life President and one of our Trustees, as the Trisomy 9 Support Group
- Now an umbrella organisation for all those affected by a rare chromosome or gene disorder associated with developmental delay (DD)/intellectual disability (ID)



Unique Membership

- Over 29,000 members
- 150-200 new families join every month
- 120 different countries



Country Name	Members
U.K.	13,209
U.S.A.	9,996
Australia	1,758
Canada	1,084
New Zealand	453
Germany	353
The Netherlands	339
Spain	227
Brazil	222
France	221

Rare Conditions



unique
UNDERSTANDING GENES
& CHROMOSOMES



Chromosomes

- We all have 23 pairs of Chromosomes. They carry the genes, with instructions telling our bodies how to grow and how to work properly.
- Under a microscope, they look something like this:



- The people Unique help all have material missing, added on or rearranged on one of their chromosomes, or a change to one of their genes. This means the instructions for growing, learning and working properly aren't the same for them

Living with a rare genetic condition

Individually rare but collectively common



1 in 17

people are affected by a rare condition at some point in their lifetime



A rare condition affects fewer than

1 in 2000



There are over

7000

rare conditions, with new conditions regularly identified through scientific progress



7 in 10

rare conditions affect children. And more than

3 in 10

children with a rare condition die before their fifth birthday



- Lack of information or understanding
- Can be life-limiting or life-threatening
- Lack of a pathway to follow
- Struggle for recognition
- Challenges finding support
- Ignorance of professionals
- Lack of evidence-based treatments
- Lack of research



The Impact of a Diagnosis

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



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

What on earth does this mean for my baby?

XXXY – it looks like a bad hand at Scrabble!

What's it called?

- 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



What Unique can Offer Families



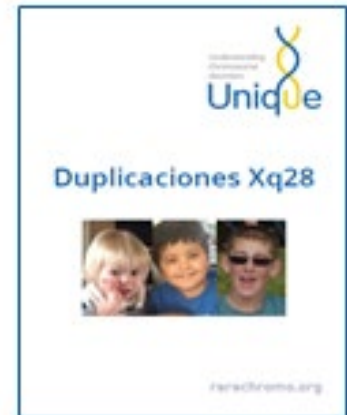
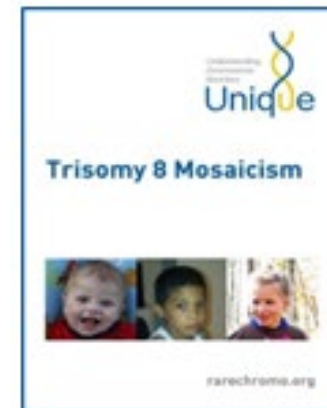
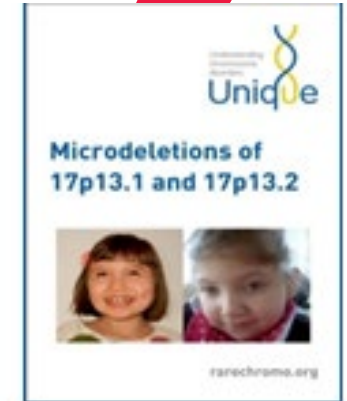
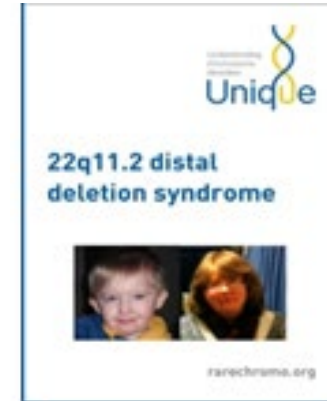
What Unique can Offer Families



Unique Guides for Families and Professionals



- Information guide project launched in 2004. Information often focused on the “worst case scenario” or was written in complex, medical terms that weren’t easily accessible to families.
- Over 250 guides to genetic conditions (FTNW, FTNP & QFN)
- Developed in collaboration with clinical and academic experts
- Over 550 translations across 22 languages



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



Practical Guides

Useful with or without a diagnosis



Understanding Chromosome & Gene Disorders
Unique

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

Understanding Chromosome & Gene Disorders
Unique

Further education, training and work

Understanding chromosome disorders
Unique

Communication

Understanding chromosome disorders
Unique

Behaviour and Sensory

Understanding chromosome disorders
Unique

Sleep problems

rarechromo.org

Understanding chromosome disorders
Unique

Feeding guide for children with rare chromosome disorders

rarechromo.org

Understanding Chromosome & Gene Disorders
Unique

Carers Wellbeing

rarechromo.org

Understanding Chromosome & Gene Disorders
Unique


Education

rarechromo.org

Other Information

EasyRead Guides

- 16p13.11 microduplications
- Deletions and microdeletions
- Duplications and microduplications
- 21q22.13 deletions
- 2p16.3 (NRXN1) deletions
- BBSOAS
- CDK13-related disorder
- DYRK1A syndrome
- Gene Variants
- Wiedemann-Steiner (WSS) syndrome




Easy read guide for people with a deletion or microdeletion

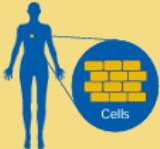
rarechromo.org

1


This leaflet is for people with a deletion or microdeletion




Before we learn about deletions and microdeletions, first let us learn a little bit about our bodies.



Our bodies are built from millions and millions of tiny building blocks called cells.



Our cells contain chromosomes.



Chromosomes are full of instructions that tell our bodies how to work. These instructions are called genes.

2



Why do I have a deletion?
Some people have a deletion because their mum or dad have it.



Some people are the first person in their family to have it.



If you had a child, they could have the same deletion too.



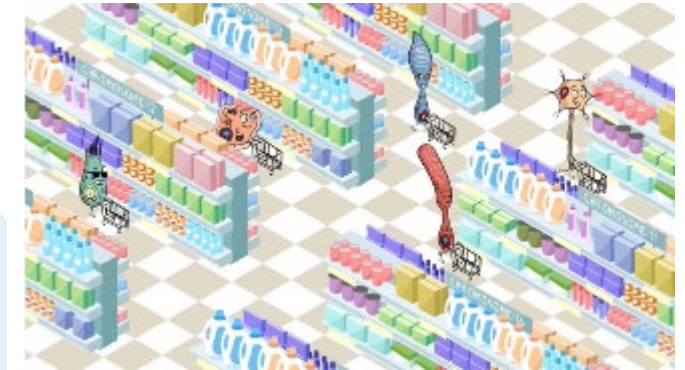
How do I know I have a deletion?
You can only find out by having a genetic test.

8

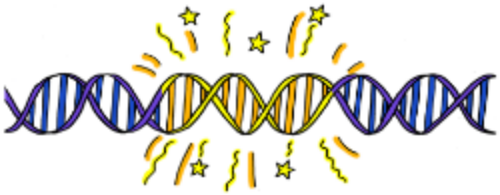
Other Information

Animations

- ArrayCGH and DNA sequencing

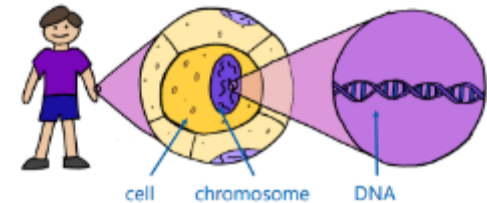


My Chromosome Story



My Chromosome Story

A picture book for chromosome 15q13.1q13.2 deletions



cell chromosome DNA

Your body is built from tiny building blocks called cells. Almost all of your cells contain DNA, which is full of instructions on how to make you. The instructions are quite long and complicated and so they are split into different parts called chromosomes. It might help to imagine the instructions are like a big book, and each chromosome is a different chapter.

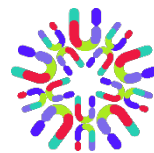
Family Days



Raising funds for Unique

- Unique relies on voluntary donations and fundraising
- Families, friends and supporters participate in a wide range of fundraising for us, from running, to baking cakes, sky diving, to dressing down!





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