Rare chromosome and gene disorders and the role of Unique in supporting families

About rare chromosome and gene disorders (RCDs)

- DNA contains genes, which tell our bodies how to develop and function. This information is organised into 23 pairs of chromosomes (46 total) of bundled up DNA.[1]
- Rare chromosome disorders can occur when all or part of a chromosome is duplicated, deleted, or re-arranged.[2]
- Rare gene disorders can occur when a change is so small only a single gene is affected.
- Individually, RCDs are reported in no more than hundreds of cases worldwide, with many being ultra-rare or even unique.
- Collectively RCDs are common, it is estimated that up to one in every 150 babies has a rare chromosome or gene disorder, with many showing symptoms from birth or early childhood.[2]
- RCDs can have a severe impact on daily life and/or development and some are life threatening, leading to stillbirth, infant or premature death, although others might present with milder symptoms.

Diagnosing rare chromosome or gene disorders

- RCDs usually present in babies or children with one or a combination of:
  - unusual physical features
  - developmental delays
  - learning disabilities
  - complex medical conditions, such as major organ defects (e.g., cardiac, lung, kidney), skeletal abnormalities (e.g., scoliosis, kyphosis), epilepsies, poor muscle tone, feeding or eating difficulties, cleft lip and palate, sight and hearing defects etc.
- They can be diagnosed in adults who have suffered repeated miscarriages, stillbirth, or undiagnosed infertility.

Given the rarity of RCDs, many families have experienced a prolonged diagnostic odyssey over many years, and are only now receiving a diagnosis in adulthood thanks to new genomic technology

- Since completion of the Human Genome Project, methods of analysing people's chromosomes and DNA, such as microarrays and DNA sequencing, have become increasingly more readily available.
- For example, the rate of people with a learning disability being diagnosed with an RCD has increased from 7% before 2008 to 40% today and continues to rise as DNA sequencing techniques and interpretation methods are refined.

About Unique

- Founded in 1984 with just five families, Unique currently have;

  over 29,000 member families, representing more than 31,000 affected individuals

  in over 120 countries worldwide.[3]
- Unique facilitate a network of families across the world living with RCDs associated with learning disability/developmental delay, bringing them together to form mutual support networks.

Registry

- Underpinning Unique's work is its world-leading database – the largest, most wide-ranging of its kind in the world which documents the impact of living with an RCDs over a lifetime.[4]
- With the rollout of genomic testing across the NHS, the number of families registered on the database has risen exponentially;

around 200 new families joining per month.

Unique have found RCD-specific matches for over 85% of member families – many of whom were told they were alone in the world; and provides networking opportunities to all member families.

Sharing unparalleled knowledge about RCDs

- Unique have produced;

over 300 family-friendly, medically verified information guides to >250 RCDs, for families, clinical geneticists, genetic counsellors, paediatricians, neurologists and other healthcare and allied professionals.

Many have been translated into 21 languages.

References

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2. Keeping you informed. Unique Website. Available at: https://rarechromo.org/information/
3. Our History. Unique Website. Available at: https://rarechromo.org/our-history/
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