

# Unique™

## Evidence-based information guides to rare chromosome disorders for families and professionals

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### Background

Unique is a world-leading support group for families with a rare chromosome disorder (RCD). At March 2010, Unique has 7250 member families with an RCD, representing more than 8300 affected individuals.

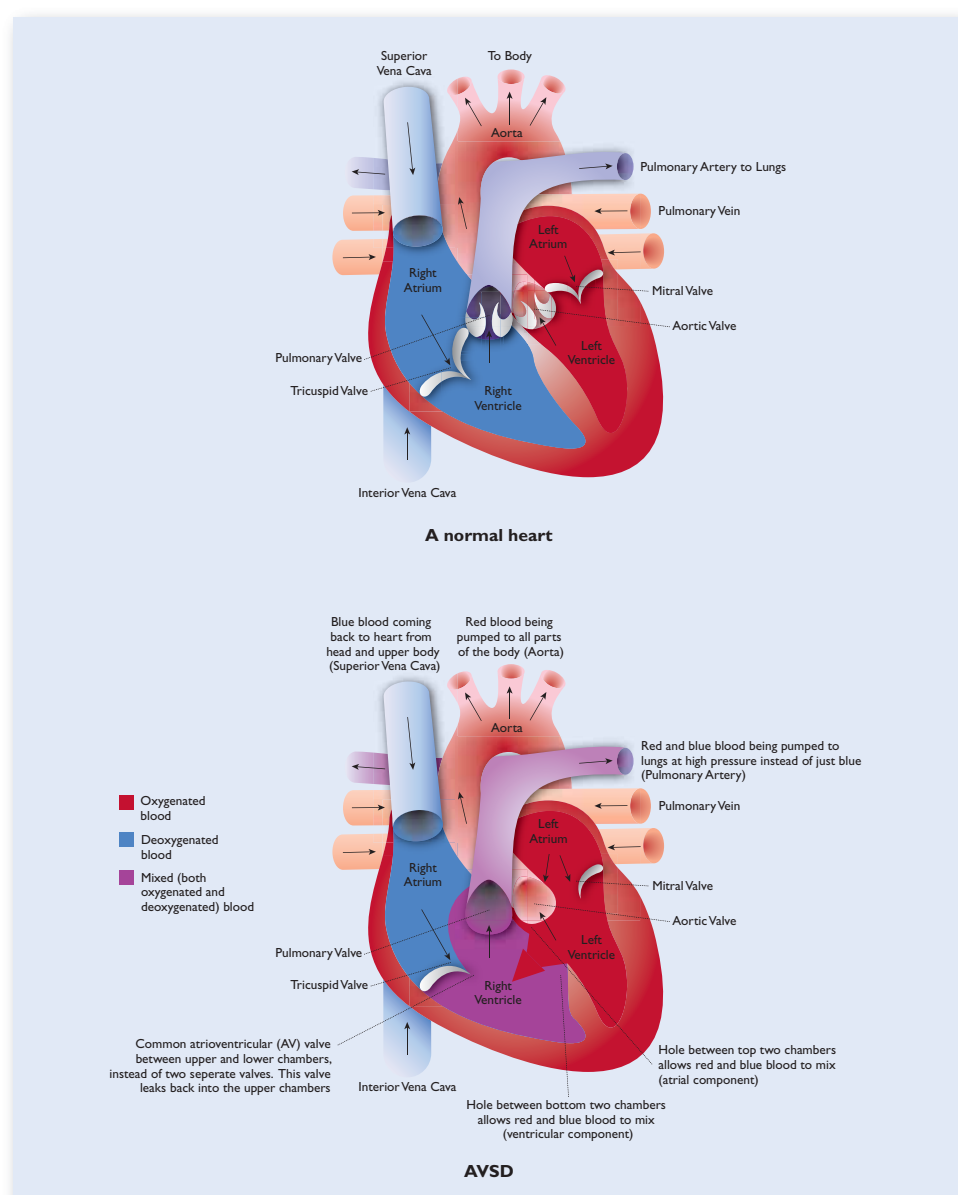
Unique generally does not work on behalf of families with a pure form of a more common chromosome disorder since support and information is available elsewhere. More common disorders include Down, Klinefelter (47,XXY), Turner, Patau, Edwards and Fragile X syndromes.

### 2 in 3 families complained of a lack of information

Families receiving a diagnosis of a rare chromosome disorder frequently experience severe distress<sup>1</sup>. This distress is

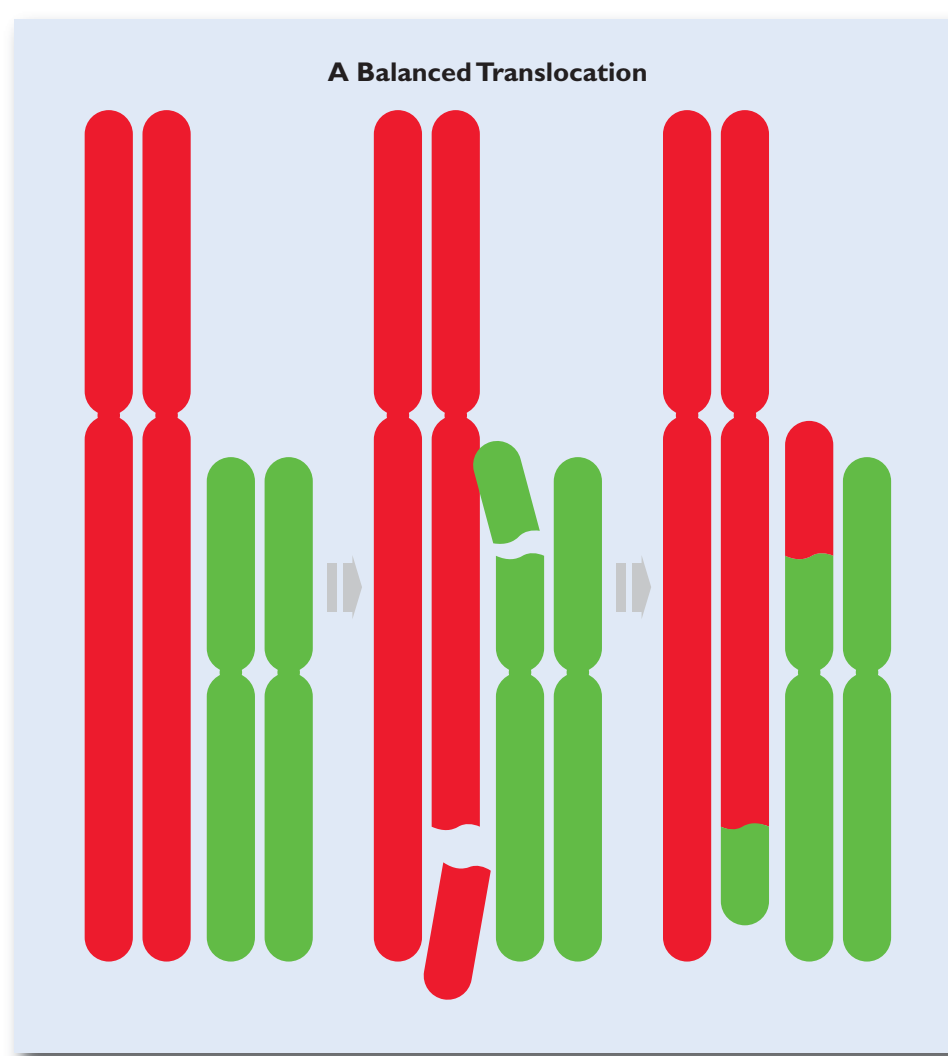
compounded by a lack of information. Two thirds of families receiving a diagnosis of an RCD in their child complained of a lack of medical information<sup>1</sup>.

Used with face-to-face information, written information relieves anxiety and stress and can improve understanding and recall<sup>2</sup>. Information guides such as leaflets are more likely to discuss a genetic issue comprehensively than individual letters<sup>3</sup>. Families can keep information guides for reference and to inform other family members, health and education professionals.



### Method

- In 2003, Unique surveyed medical information on RCDs suitable for families. They found information on more common chromosome disorders developed by the genetics department at Guy's Hospital, London and by Contact a Family. The Genetic Interest Group (GIG) provides some generic information on chromosome disorders, developed with Eurogentest. The published information available was relevant to six per cent of member families (413). The number of members with any particular disorder ranges from one to 259 (47,XXY).
- To bridge this information gap, Unique embarked on a pilot project to publish a range of family-friendly disorder-specific information guides. The guides were also developed as a resource for health and education professionals.
- A working list of disorders was compiled, prioritising the most common disorders faced by Unique families and those where no other family-friendly information was available.
- Unique surveyed families' information needs by asking them what they most wanted to know and who gave the most helpful answers. Unique asked what questions members still had unanswered.



- The medical literature was searched and key features established. Any current research was identified. Medical or genetics professionals were found with a current or recent interest in the disorder.

### Unique has published more than 100 verified information guides

where possible substantiated by medical and educational reports. Parental reports can be reliable sources of information on individual development<sup>1</sup>. Unique's experience is that information from other families is particularly valued by families.

- Unique drew up disorder-specific questionnaires targeting families' most frequently expressed information needs. Questions differ from those in a standard medical history because parents' perceptions of a child's quality of life are different from health professionals' perceptions<sup>4</sup>. Unique surveyed members on: pregnancy and birth; feeding; neonatal experience; growth; birth defects; medical and dental disorders; vision and hearing; puberty; cognitive and motor development; education; sleep; behaviour and mental health.
- Additional questionnaires went to families of adults to elicit information on puberty, transition from children's to adult services, social wellbeing, social integration, progression or development of any medical conditions, mental health, learning outcomes, work and work experience and support services outside the family.
- Families were invited to supply photographs. Permissions to publish any photographs were sought.

### Outcome

Unique surveys elicited high response rates of up to 100 per cent. Compilation of results is ongoing. A high proportion of families said that Unique was their most helpful source of information on their child's chromosome disorder.

### Development of disorder-specific information guides

- Information from the surveys, database and medical literature is collated in tables. A text is drafted that answers families' most pressing questions. Texts contain both factual information and accounts of family experiences<sup>3</sup>.
- Illustrated information guides are reviewed by families, medical and genetics professionals and Unique's chief medical adviser Professor Maj Hultén.
- Information guides are freely available from Unique at [www.rarechromo.org](http://www.rarechromo.org).

- As new information emerges, Unique updates its information guides.

Unique has published more than 100 guides. Information guides cover the diagnoses of 68 per cent of Unique members. Seventeen information guides have been translated and translations verified by geneticists. Translations are available in German, French, Spanish, Italian and Dutch. Health professionals are informed about the information guides at relevant conferences, meetings and events.

### Further work

Unique is continuing to publish and update RCD-specific information guides, including newly-emerging microdeletion and microduplication syndromes, as well as more generic guides on balanced reciprocal translocations, Robertsonian translocations and sSMCs and on technological advances such as arrayCGH (microarray). Translations are continuing.

**EuroGentest** The support of Eurogentest for some of this work is gratefully acknowledged.

### References

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### Before: Information was helpful to 6% of families



### After: Information is now helpful to 68% of families

[www.rarechromo.org](http://www.rarechromo.org)