Guide Updates
Genetics is a very fast-moving and ever-evolving field so our guides inevitably require periodic updates. However, with our limited resources, we are not able to update each guide as often as we would like. Some guides are updated with information from the medical literature, while others also involve a member survey. This depends on the significance of the new information and how many members we have with a specific disorder. We are also currently liaising with many professionals worldwide who have published research findings in medical journals regarding specific genetic changes; some have already voluntarily dedicated their time to help update a few of our existing guides. We are hopeful that we will be able to update more guides in the future with the help of our professional volunteers who are also leading the way in genetic disorder research.

If you are a professional with a keen interest in a specific genetic change and would like to help write, update or review a Unique guide, please don’t hesitate to contact us (info@rarechromo.org).

Translations
We often receive requests from families and genetic clinicians and counsellors worldwide for a guide to be translated into another language so that it can be shared with non-English speaking families and professionals. Our translators all work on a voluntary basis, and many are doctors or medical professionals who work closely with families affected by rare genetic disorders.

If you are a professional with a scientific or medical background and would like to help translate a guide, please let us know (info@rarechromo.org).

Inform Network Support

Rare Chromosome Disorder Support Group,
The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK
Tel: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Join Unique for family links, information and support.
Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at: www.rarechromo.org/donate
Please help us to help you!

Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and Unique does its best to keep abreast of changing information and to review its published guides as needed. This leaflet was written by Unique (AP)
2020 Version 1 (AP)

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Unique Guides

*Unique* has been writing information guides for families since 2003. We are very happy to be able to share so much information, completely free to families and professionals worldwide:

We currently publish over 500 guides on our website, they include:

- 185 rare chromosome disorders guides
- 48 single gene disorders guides
- 24 guides for specific genetic topics
- 9 study/day/weekend reports
- 271 translations in 14 different languages
- 22 practical guides

(February 2020)

**Disorder specific information guides**

The *Unique* genetic disorders guide team consists of two part-time information officers. Their job is to research and write new guides, update existing guides, edit and format guides written by other professionals and format translations. They also write, edit and format information guides explaining other genetic topics.

Guides written by *Unique* are reviewed on a voluntary basis by clinical geneticists and other professionals working in this field. Most of our single gene disorder guides are written by clinical geneticists who kindly volunteer their time to help share their professional knowledge of newly identified disorders. Translations are written and verified by dedicated volunteers, most of whom are also professionals involved with the care of children and families affected by rare genetic disorders.

We strive to present guide content in a family-friendly way with accurate information which includes that from our members, those who know the most about living with rare genetic disorders. We cannot emphasise enough how important it is for families to be involved in guide production.

Many different factors are taken into consideration when selecting guide priority, including the number of *Unique* members, the amount of information available in research articles and the availability of expert writers and reviewers.

**Aim**

We produce information guides in order to share as much knowledge as we can about specific genetic disorders. We include information about chromosomes and genes and what genetic test results mean. We also detail possible developmental and medical features for children and adults, together with therapies and/or treatments; some of our more recent guides also include management plans.

The insights and experiences that parents kindly share about their children of all ages with a specific genetic disorder can be of great help and reassurance to other parents, as well as to clinicians counselling families.

Concerns and advice on many topics such as behaviour, education and social issues are shared.

*Unique* guides are designed to help families affected by rare genetic disorders but can also be of help when explaining to schools and social workers about a child's specific strengths and difficulties. They can help explain more to extended family members and friends, as well as medical professionals who are involved with the care of a child but do not have much knowledge of their specific genetic disorder.

**Rare chromosome disorder guides**

When writing a new guide, we make a thorough assessment of recent research findings and publications in medical journals. During this part of the process we attempt to identify a suitable reviewer for the completed guide, an expert in the genetic disorder described, if possible.

We then formulate a detailed survey to send to *Unique* members, so they can share their invaluable information about the developmental and medical difficulties and achievements of their child(ren). We also gather further information from our secure offline database, details that families have already shared. We are always incredibly appreciative when families share comments and photos, these personal insights really help newly diagnosed families to better understand the reality of living with a specific genetic disorder. We do request that anyone willing to complete a survey is also a member of *Unique*. This is not only so that we can store information securely, there are many other benefits to being a *Unique* member!

When the background information has been read, and the bulk of the scientific data has been assessed, we can then compare and add the information that families have kindly shared (most of which is not found in research articles published in the medical literature). We can then start writing the guide.

For those of you who have already been part of this process, we do send a few reminder emails about survey completion. We really do appreciate you are especially busy, but your information makes so much difference to the quality of *Unique* guides and to the lives of newly-diagnosed families. We are trying to collect as much information as possible to make the guide as informative as we can and help extend knowledge, understanding and care management planning.

The completion rate to our surveys varies from guide to guide but we are always grateful for any information parents feel comfortable sharing. The surveys are often quite lengthy, so if we do ask you if you’d be willing to complete a survey and you feel you can only complete part of it, then please do, as every detail can help.

Once the first draft of the guide is complete, we send it to a carefully selected medical/scientific professional to review. Reviews are carried out on a voluntary basis and such specialists are also incredibly busy. Subsequent drafts are then drawn up until we are all happy with the contents. We send a copy of the ‘almost final’ version to families who have completed a survey and/or sent comments and/or photos so they can see how their information has been presented. Following any alterations recommended by *Unique* members, the guide is finally ready to be placed on our website.

**Single gene disorder guides**

Most of our single gene disorder guides are written by clinical geneticists who are involved in the care of families with specific gene variants. These guides do not commonly involve a survey since we do not generally have enough relevant family members. However, a few of these guides have been written by us, and a few have involved surveys. We have a long list of single gene disorder guides in progress, and an even longer list of guides we would like to produce. However, we are only able to include autosomal dominant rare single gene disorders associated with developmental delay and learning disability.