

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 world wide for a guide to be translated into another language so 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).

Unique's first guide was published in 2003 shortly after the employment of our first guide writing information officer, Prisca Middlemiss. Prisca was a member of *Unique* and a well-established medical journalist. She understood the complexities of genetic disorders on both a personal and a professional level. Prisca wrote numerous information guides and helped countless families over a 13 year period. She worked tirelessly and with great dedication despite her fragile health. Prisca passed away in 2016 and leaves an extraordinary legacy in the form of *Unique* information guides, a legacy that we are determined to continue.

The relatively recent advances in genetic technologies has resulted in an ever-increasing number of children and adults being diagnosed with a rare chromosome and/or gene disorder. For a great number of these genetic changes, there is very little information available for families. We have an extensive list of new guides we would like to write, and as time goes by, the list of older guides that require updating lengthens. We are however limited in what we can achieve with guide production, given our tiny team and budget. In the meantime, please know that, as *Unique* members, you are not alone. If possible, we will put you in contact with other members who share the same or similar genetic change. We also have a private Facebook café where members can ask each other for advice and share information. You can also contact us should you have any specific questions and we will try to answer them in a timely manner as best we can.

Inform Network Support



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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)

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Understanding Chromosome & Gene Disorders

Rare Chromosome & Single Gene Disorder Information Guide Production

rarechromo.org

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 almost 700 guides on our website, they include:

- 190 rare chromosome disorders guides
- 61 single gene disorders guides
- 24 guides for specific genetic topics
- 9 study day/weekend reports
- 369 translations in 16 different languages
- 26 practical guides

[July 2021]

Writing 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 (RCD) guides

These guides cover many different types of chromosomal changes including copy number variants (CNVs). 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 all matters relating to their child's health, development, behaviour, education, strengths and difficulties. We also gather further important 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 (SGD) 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 rare single gene disorders associated with developmental delay and learning disability.