RARE
resources
SCOTLAND

A guide for families with a child or young person with a rare, genetic or undiagnosed condition

Who is this guide for?

This guide is for people who care for a child or young person who has, or may have, a genetic, rare or undiagnosed condition.

This guide provides a brief introduction to what it means to have a genetic, rare or undiagnosed condition and useful links to where you can find sources of reliable information and support.

The information in this guide has been produced by Genetic Alliance UK, in collaboration with families in Scotland.

Top Tips

Rare Resources Scotland is a downloadable toolkit of information and sources of support for people living with rare, genetic and undiagnosed conditions in Scotland.

geneticalliance.org.uk/support-and-information/rare-resources/

What are genetic, rare and undiagnosed conditions?

Genetic, rare and undiagnosed conditions cover a broad range of health conditions that will also vary in their severity and the impact they have.

There are many types of genetic and rare conditions and how a condition affects a family will depend on a number of factors. However, it is not uncommon for families of children and young people with rare, genetic and undiagnosed conditions to experience similar challenges.

These often include:

- A long journey to diagnosis
- Uncertainty about the future
- Lack of understanding from professionals involved in the care of their child
- Poorly coordinated care with lots of appointments and different healthcare professionals involved
- Difficulties in accessing information and support from an expert, or patient group, for their child's condition
- Experiencing feelings of isolation

A genetic condition is caused by changes in an individual's DNA sequence. We all have lots of changes in our DNA sequence and often these don't have an impact. Sometimes, however, these changes can result in learning disabilities, developmental delay and other health problems. Genetic conditions may be single gene, chromosomal or complex disorders.

Genetic Alliance UK provides information on genetic conditions on their website - geneticalliance.org.uk

A rare condition is any condition that affects less than 5 in 10,000 people. There are between 6,000 and 8,000 rare conditions.

Rare Disease UK provides information on rare conditions on their website - raredisease.org.uk

Sometimes a child or young person may be suspected to have a genetic condition, but genetic testing has so far failed to identify the change in the DNA that has caused it. Sometimes undiagnosed conditions are called 'syndromes without a name' or 'SWAN' for short. Children affected by a syndrome without a name can have a range of different symptoms and each child is likely to be affected differently.

SWAN UK is the only dedicated support network available for families of children and young adults with undiagnosed genetic conditions in the UK - undiagnosed.org.uk

Where can I find reliable information?

When your child receives a diagnosis of a genetic and/or rare condition, you may find that there is not much information available.

Your GP or specialist clinician should provide you with information at the time of diagnosis, even if there is very little information available. If information is not provided, don't be afraid to ask where you can provide reliable information.

You may find these organisations helpful in assisting you to find good quality, reliable information:

Genetic Alliance UK is a national alliance of organisations with a membership of over 200 charities that support children, families and individuals affected by genetic conditions. You can search a list of member organisations on the Genetic Alliance UK website.
0300 124 0441
www.geneticalliance.org.uk

Contact is a charity for families with disabled children. Their website provides a searchable A to Z directory of medical conditions with links to information and support organisations.
0808 808 3555
https://contact.org.uk

Unique provides support for families affected by rare chromosome disorders or autosomal dominant gene disorders. They provide free guides on specific chromosome and gene disorders.
0188 372 3356
www.rarechromo.org

Orphanet is an online portal that provides information on individual conditions.
www.orpha.net

Where can I find support?

It can be comforting and helpful to talk to someone with, or who has a child with, the same condition – someone who has experienced the same things you are facing.

Having a rare, genetic condition can be isolating because there aren't many other people living with the condition. The same goes for being a parent, carer or family member to someone with a genetic, rare or undiagnosed condition.

Genetic Alliance UK can help you find support. On the Genetic Alliance UK website, you can use the search function on the members' page to find groups that may be relevant to you in the UK. bit.ly/geneticallianceuk

Sometimes, particularly in the case of very rare conditions, it may not be possible to find an organisation specifically for your child's condition. This does not mean that there is not support and you may wish to consider Umbrella Organisations.

Umbrella organisations

Often conditions fall within wider categories of conditions. For example, Danon disease is extremely rare and doesn't have a specific support group. But Danon disease is a metabolic condition, so people can access support and information from the umbrella organisation Metabolic Support UK. If you want to know what type of category your child or young adult's condition falls under, it is a good idea to ask your health professional.

Online Forums

The internet is home to a wealth of virtual areas for people to meet and talk about their experiences with a rare, genetic or undiagnosed condition.

Contact Online Community is an online forum set up by the organisation Contact for parents of children with disabilities. bit.ly/rrcontactonline

Rare Resources Guide

The Rare Resources guides have been produced by Genetic Alliance UK for families who have recently received a diagnosis of a genetic or rare condition, are on the journey to a diagnosis or who have been told their child's condition is so rare they might not get a diagnosis.

The guides contain a wide range of general information on rare, genetic and undiagnosed conditions as well as information on how to access reliable information, care and support.

Rare Resources contains the following guides:

- Genetic, rare and undiagnosed conditions explained
- The journey to diagnosis
- Support and information for parents and carers
- Support and information for your child
- Using the NHS in Scotland
- Information directory detailing support services available in Scotland

The Rare Resources guides can be downloaded from bit.ly/rrgeneticallianceuk

To request a hard copy, please contact
Genetic Alliance UK
0300 124 0441
contactus@geneticalliance.org.uk

Top Tips

From Families, for Families

Find reliable information and support

It may be helpful to find information on your child's condition. For some conditions there may be a support group available. The Rare Resources guides provide information on how to access reliable information and support, even if your child does not have a diagnosis.

Don't be afraid to ask for help

Remember it is completely normal to feel overwhelmed from time to time. Don't be afraid to ask for help if you need it.

Meet other parent carers

It can be very beneficial to meet others who are experiencing, or have experienced what you are going through.

Understand your rights, and the rights of your child

You may find that you and your child are eligible for practical, emotional and financial support. The Rare Resources guides contain information on the support available to families in Scotland.

Get organised

Keep a copy of all letters and information relating to your child's health together. This will ensure you have the important information together and help you keep track of your child's progress.

Your **Notes**

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We acknowledge the support of the Scottish Government through a CYPFEIF and ALEC Fund Grant in producing these Rare Resources. Produced March 2019. Reviewed March 2021