RARE resources SCOTLAND

2 The journey to diagnosis

RARE resources SCOTLAND

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

The Rare Resources guides have been developed in collaboration between Genetic Alliance UK and families in Scotland. The guides provide links to reliable sources of information and support, and contain 'top tips' from other families.

Rare Resources contains the following guides:

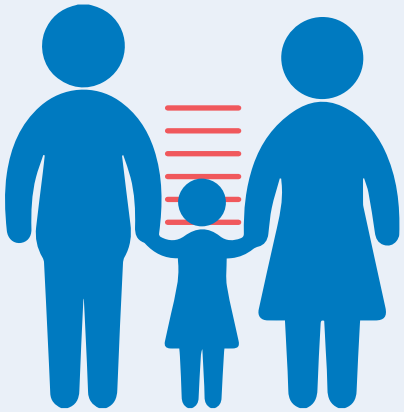
1. Genetic, rare and undiagnosed conditions explained
2. The journey to diagnosis
3. Using NHS Scotland
4. Support and information for parents and carers
5. Support and information for your child
6. Information directory detailing support services available in Scotland

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

If you have an questions about our Rare Resources guides, please contact us on 0300 124 0441 or contactus@geneticalliance.org.uk

GENETIC ALLIANCE UK

2.1 What to do if you are worried about your child's development?



If you have concerns about your child's development, it is a good idea to discuss this with your health visitor or GP.

Your health visitor or GP should listen and then discuss your concerns with you, examine your child and consider options. If they are unable to offer a diagnosis, they may refer you to other health professionals for their opinion.

Your GP may refer your child to a paediatrician for their opinion. A paediatrician is a doctor who has special training in medical care for babies and children.

If your GP or paediatrician suspects a particular condition, or a particular type of condition, your child may be referred to a specialist clinician.

Your GP may also refer you to a nurse or a healthcare professional who can help with particular symptoms. For example, a child experiencing muscle weakness may be referred to a physiotherapist. Nurses and health professionals may be able to help you obtain a diagnosis.

If a genetic condition is suspected by your GP, paediatrician or specialist clinician, a referral to an NHS genetics service will be arranged for you. You can also ask for this referral to be made.

It may take a long time to find a diagnosis and for some families, and it may be that diagnosis is not possible. However, there is support available to families, at all stages of their journey.

2.2 How to access genetic testing for your child

To access genetic testing you must contact your GP or specialist clinician who will arrange a referral for you to an NHS Regional Genetics Service.

Here we explain what will happen at that appointment, and how long you might have to wait for a result.

Genetic testing on the NHS is available through your specialist clinician who orders the tests that are appropriate for you from an NHS-certified laboratory.

The clinician will request a test only if they know that the results will help them provide you with the most appropriate healthcare.

NHS policies define who is most likely to benefit from specific genetic tests. Your specialist clinician will collect your sample for genetic analysis and send it to the laboratory. The laboratory will then analyse and interpret the results.

Your clinician will then be available to talk you through what your results mean. Anyone having a genetic test on the NHS is also likely to see a genetic counsellor.

If the test will be looking to determine whether you are affected by a serious genetic condition, counselling will be available both before and after you take the test.

The British Society for Genetic Medicine website has information on genetic services. bit.ly/rrinformationforpatientsandfamilies

What is genetic counselling?

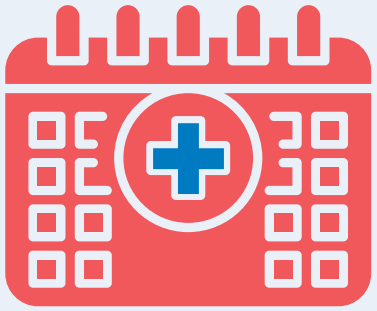
Genetic counselling is a service that provides support, information and advice about genetic conditions. Genetic counsellors are healthcare professionals who have received training in the science of human genetics (a genetic counsellor or a clinical geneticist). What happens during genetic counselling will depend on exactly why you've been referred.

It may involve:

- Learning about a health condition that runs in your family, how it's Inherited, and which family members may be affected
- An assessment of the risk of you and your partner passing an inherited condition on to your child
- A look at the medical history of your family or your partner's family and drawing up a family tree
- Support and advice if you have a child affected by an inherited condition and you want to have another child
- A discussion about genetic tests, which can be arranged if appropriate, including the risks, benefits and limitations of genetic testing
- Help understanding the results of genetic tests and what they mean
- Information about relevant support groups
- You'll be given clear, accurate information so you can decide what's best for you

Your **Notes**

2.3 What happens at a genetic testing appointment?



After being referred to an NHS regional genetics service you will usually be sent a leaflet or letter explaining what will happen when you attend the clinic for your appointment. This letter often asks you to bring your child with you to the appointment.

Usually they will write to you after the appointment summarising what was discussed so you have a written record of it.

If you are a single parent or your partner cannot attend the appointment with you, you may wish to take along a family member or trusted friend for support. They may also remember points about the appointment that you miss.

Often at the start of the first appointment, the geneticist will ask parents what they are hoping to find out, for example:

- A diagnosis for their child.
- How their child will be affected in the future.
- If any future children could be affected.
- Whether other children in the wider family could be affected.

Most of the appointment will be spent going through the 'genetic assessment process'. This is a diagnostic process where clinical geneticists work with laboratory staff and other medical professionals to explore the possible reasons for the different developmental patterns a child has.

Their aim is to identify a genetic syndrome or condition that explains the child's difficulties.

The cause of the condition might be a single-gene disorder, or it might be that a particular chromosome has a piece missing (deletion) or an extra piece (duplication), or it may be that pieces of chromosomes have 'swapped around' (translocation).

Identifying the genetic cause of a child's health problems occurs through a combination of investigations:

- Asking about the child's development in detail.
- Asking about the child's family medical history.
- Physical examination including photographs.
- Laboratory investigations (if necessary, blood samples are taken during the appointment or shortly afterwards).

Asking about the child's development in detail

The history of a child's development and the pattern of problems that they currently have or have had in the past is a key component of the assessment.

It can be frustrating having to repeat your child's history to yet another professional, but this is a very important part of the assessment. This can sometimes feel upsetting as you have to focus on all the things that make your child different or that they can't do.

Asking about the child's family medical history

The geneticist will ask about your family history to see if they can identify patterns that might give clues to the cause of your child's problem.

Family history is important because if the problem is genetic, then it may be the result of changes in genes or chromosomes that were passed down from one or both of the parents. Sometimes it may be possible to make a specific diagnosis based on these historical descriptions alone without the need for genetic testing.

You might find it difficult to describe your family history due to adoption or having little contact with family.

Physical examination including photographs

The aim of a physical examination is to identify any particular physical features that might give the geneticist a clue about a possible diagnosis.

This can be an odd, and sometimes upsetting, process for both the child and parents.

Laboratory investigations

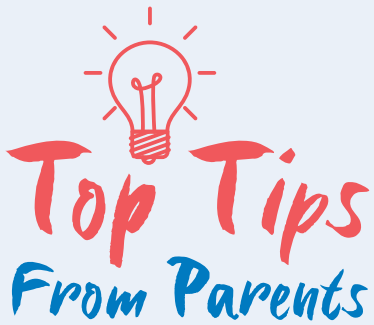
Laboratory investigations are usually the final part of the diagnostic process. Once a geneticist has an idea about a potential diagnosis for the child, they may order laboratory tests straight away to confirm the suspected diagnosis.

Samples of the child's DNA (and sometimes the parents') will be taken to test and look for changes in the DNA. The samples needed can vary and may include blood, saliva or skin.

The sample will be sent to the laboratory, which will then analyse and interpret the results.

Once a geneticist has an idea about what might be a potential diagnosis for your child, they may order laboratory tests straight away to confirm the suspected diagnosis. There can often be a long wait for results.

Learn more about genetic testing by visiting Genetic Alliance UK website. bit.ly/geneticallianceuk



Before attending appointments, it is a good idea to write down any questions that you want to ask when you are there and take a notebook along.

You might also find it helpful to take a notebook along to the appointment. Some families record the appointment on their phone so that they can listen again afterwards. You should always ask permission from the other people at the appointment before recording it.

If you are worried you won't remember some of the details at the appointment you can ask your geneticist to write them down for you to look at again when you get home.

2.4 How long does it take to get results from genetic testing?

Sometimes it is not possible to undertake genetic testing straight away. This might be because your child is too young for their physical features to give any clear 'clue' about what the underlying condition may be.

In these situations, you will usually be invited back for follow-up appointments several years later when there may be more clues because your child has developed, new medical knowledge has come to light, or a new test has become available. This period is usually referred to as 'watchful waiting'.

It can be frustrating and worrying waiting for answers and many families wonder if they have been forgotten. Whilst they are waiting, many families start searching the internet to see if they can find answers.

Although this is understandable, internet searches for symptoms often produce photographs of children with a range of different conditions which can vary in the way they affect different children.

It is often the 'worst-case scenario' images that pop up and some of these images can be very upsetting for families to see.

Top Tips From Parents

Genetic testing can take a long time and you may have to wait several years for any result. there are organisations that can support you while you wait.

SWAN UK (syndromes without a name) is the only dedicated support network available for families of children and young adults with undiagnosed genetic conditions in the UK. bit.ly/rrjoinswanuk

2.5

Preparing for appointments

You may have to attend a number of appointments with a number of different health professionals on your journey to a diagnosis for your child. It is normal to feel overwhelmed and anxious about attending appointments.

Top Tips From Parents

Families tell us that preparing for appointments can sometimes help with these feelings.

It is a good idea to prepare a list of things that you would like to discuss during your appointment.

Your list can include things that you want to tell your GP or specialist clinician - for example, new symptoms your child has had or any changes in their patterns of behaviour.

Some families find it helpful to make videos to take along to show their child's GP or specialist clinician, for example if you think that your child might be having a seizure or is displaying unusual behaviours.

It is also a good idea to write down any questions that you would like to ask.

You might find it helpful to attend with a partner, a family member, friend or advocate. They can offer support, but also remind you to ask a particular question or write notes for you.

It is a good idea to keep details of your appointments, letters and test results together. This will allow you to keep an up to date record of your child's journey to diagnosis.

2.6 What does getting a diagnosis mean?

For most families, getting a diagnosis remains just as important to them as their child grows up. Without one, families can struggle to access the right support. Not having a diagnosis may mean not knowing what the future holds for their child or if other children they may have in the future could be affected.

'Wanting a diagnosis is not about expecting a cure or a magic wand... but knowledge is power.'
SWAN UK member

Although parents generally know that having a diagnosis will not significantly change their child's life, they hope it will give them a greater sense of what to expect.

Families of children who have been given a genetic diagnosis of a very rare chromosome condition, where the child may be one of only a few in the world known with that condition, can sometimes feel disappointed with the information given to them.

Families generally hope that a diagnosis will give them an indication as to what the future holds for their child but this is not always the case.

Although families are usually glad they can now explain what is wrong with their child, the technical description of the genetic change in their child's genes or chromosomes does not give them much idea of what to expect from the future. If their child is the only one, or one of a few in the world with this known genetic change, they can still feel quite isolated.

'I wasn't expecting a cure, I knew a genetic condition was lifelong, but I was expecting an understanding of what it meant for my son's life. What I got was a string of letters and numbers.'
SWAN UK member

However, while these rare diagnoses do not always tell the family much about what they can expect in their child's future, it does at least give them a reason for their child's conditions - even if it is unlikely to be understood by many people outside of the field of genetics.

'I am regularly caught between not letting her diagnosis define her life and also raising awareness of rare conditions and what they mean

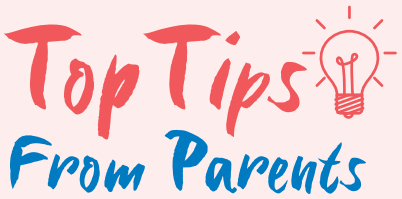
for families like ours. I often say that although I wish I could take away H's challenges I wouldn't take away her wonky genes. They are so intrinsically her that to change her would probably completely change her, the person we love.'
Parent of a child with a rare condition

Many families say that without a diagnosis they feel like they are treated like a 'neurotic mother' or told that their child will 'catch up' or 'grow out of it'. Having a diagnosis gives them a way to respond to this kind of comment.

Having a diagnosis is really important to families on a practical and emotional level.

Here are some of the main reasons for needing a diagnosis:

- **To have a sense of what the future holds**
- **Having a diagnosis could help identify potential treatments or health issues that need to be monitored in the future.** Without a diagnosis it is hard to know whether new symptoms are something to be worried about.
- **To have the child or young person's needs taken seriously.**
- **To access service and support.** Families often find it hard to access services without a diagnosis. Forms or assessment criteria have a box that asks for the child's diagnosis and families struggle to know what to write.
- **To know if other children in the family will be affected.** Without a diagnosis it is impossible to know if the condition is inherited or just a one-off. This means families don't know if future children will be affected by the same unknown condition. This affects not only the parents of the undiagnosed child, but also their siblings, aunts, uncles and cousins.



The support group Unique offers information and support to families with rare chromosome disorders and, where possible, link together families whose children have the same rare condition. bit.ly/rrunique

Your **Notes**

2.7 Why are some conditions difficult to diagnose?

The long and difficult journey many families have to go through to get a confirmed diagnosis is often called the 'diagnostic odyssey'.

There are three main reasons why a child's genetic condition remains undiagnosed:

- **It is the 'rarest of the rare'** - a condition that hasn't been seen before and therefore isn't tested for.

- **It is an unusual presentation of a known condition** - the child's symptoms might be different to those of other children with the same condition. The condition is therefore not tested for because it doesn't appear to be that condition.
- **Genetic changes are found which are of 'unknown clinical significance'.** This means that changes are found in the child's chromosomes but these are not thought to be the cause of their difficulties. Sometimes this is because the change has been inherited from one of their parents who is unaffected by the condition, or because, although the genetic material has moved around, it all appears to be there.

2.8 Accessing support for your family on your journey to diagnosis

Trying to obtain a diagnosis for your child can be a worrying time. There are people in your life, health professionals and organisations that can help you.

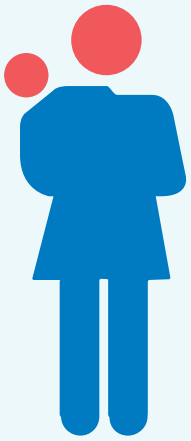
Your GP, health visitor or specialist clinician can talk you through the journey to diagnosis.

They can provide you with information on the process and information on where to seek support.

You may be in touch with a genetic counsellor. A genetic counsellor can work directly with your family to offer genetic information and will support you to make decisions for your child.

There are also parent groups and support organisations that can provide emotional and practical support for you and your family. You can find out more about how to find support organisations later in this guide.

2.9 Coming to terms with a genetic, rare or undiagnosed condition



Wherever you are on your journey to diagnosis, coming to terms with the fact that your child has a health condition can be difficult.

The news that your child has, or may have a genetic, rare or undiagnosed condition can lead to a wide range of emotions.

'Things felt less scary once we got a diagnosis.'
Workshop participant

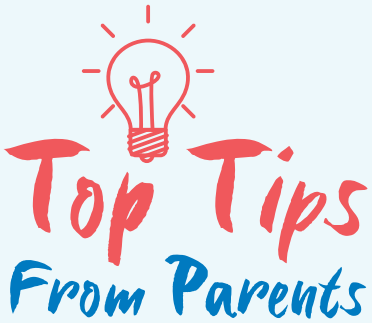
Every person deals with diagnosis, or the news that their child may remain undiagnosed, differently. There is no right or wrong way.

'We still don't have a diagnosis for my child, and it's unlikely we will get one soon. I've learnt to just focus on what my child needs now and enjoying our life together.'
Workshop participant

Some parents react to the news in a similar way to having a bereavement, others cope by learning all they can about the condition or focusing their attention on their child's specific needs.

'Looking back, I guess I went into a state of grief when we got a diagnosis.'
Workshop participant

Sometimes you might want to speak to someone about how you are feeling. You can contact your GP, your local carers centre or local support group for advice on the help and counselling they may be able to offer.



Take your time. You don't need to ask every question or learn everything about the condition all at once. Take your time to process information at your own pace and think about your feelings.

Talk. Talk to someone you are close to about the diagnosis and how you are feeling. If you don't feel that you can speak to someone you know, try an organisation that provides a support helpline (find details in the Information Directory).

Find a support group. For some conditions there may be a support group available. Support groups can provide information on a condition and support for your family.

Research. For some people, it can be helpful to find information on and research the condition.

Speak to the other families. For some people, speaking to other families who have a child with a similar condition can be reassuring and helpful.

Be positive. Remember to focus on the good things about life with your child. It can be easy to become overwhelmed by a diagnosis, so spend time focusing on positives.

Your **Notes**

2.10 What next?

I have been told my child has a rare genetic condition - what do I do now?

By having a diagnosis and knowing the name of the condition, you may be able to access information and support to help you understand the condition and how it will affect your child.

A diagnosis of a very rare condition can be frustrating.

There may be little information on the condition available and little understanding of the condition amongst health professionals and care providers. Sometimes many questions can remain unanswered.

Top Tips From Parents

You can find further information on how to deal with a diagnosis and where to access information and support in the **4 Rare Resources: Support and information for parents and carers** and **5 Rare Resources: Support and information for your child**

I have been told my child may never get a diagnosis

A significant number of children having genetic testing will not receive a diagnosis. This can be very difficult and it is important that you have support.

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

SWAN UK supports a community of families and children affected by undiagnosed genetic conditions and provides a range of information for families.

Top Tips From Parents

You can find further information on where to access information and support in the **4 Rare Resources: Support and information for parents and carers** and **5 Rare Resources: Support and information for your child**

2.11 After diagnosis – finding information and support

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.

They may be able to provide you with a leaflet, a printout from the internet, or they may be able to sign post you to appropriate information.

If information is not provided, don't be afraid to ask your GP or specialist clinician where you can find reliable information.

Your local Patient Advice and Support Service (PASS) may also be able to help you.

Patient groups often provide a valuable source of condition-specific information.

2.12 Where can I find a support group?

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 thing that you are facing.

Having a rare condition (which most genetic conditions are) can be extremely isolating because there aren't many other people out there 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

We 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.

You can also try an internet search, for example, it may be useful to you to search the name of your condition and the words 'patient support UK' in an internet search engine – be careful though as it is easy to scare yourself as often the results you find might be the worst case scenario.

Sometimes, particularly in the case of very rare conditions, it may not be possible to find an organisation specifically for your child or young adult's condition.

This does not mean there is no support available and you may wish to consider umbrella organisations and online forums.

Umbrella organisations

Often conditions fall into wider categories. For example, Danon disease is extremely rare and therefore does not have a condition-specific support group. But Danon disease is a metabolic condition, so support and information is available from an umbrella organisation such as CLIMB (Children living with Inherited Metabolic Conditions).

Top Tips From Parents

If you want to know what category your child or young adult's condition falls under, you can ask your health professional.

Online Forums

Sometimes families struggle to find a specific support group. However, the internet is home to a wealth of virtual areas for people to meet and talk about their experiences with rare condition.

Facebook

As a free informal platform, Facebook is home to a number of online support groups. You don't need to have a Facebook account to search for groups, although if you do find a relevant group you will need to sign up to join and talk to people.

If there isn't already a Facebook group for the condition, you can always set one up for the next person searching to find. It can be a great way to connect with others directly.

Contact Online Community

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

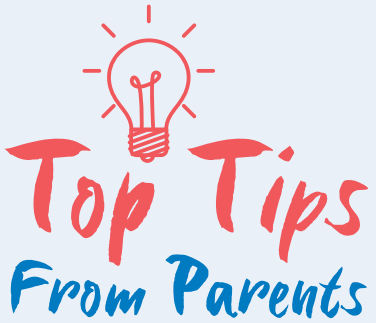
Just enter your condition into the search function on the website and you'll be able to see how many people have registered under the condition name.

Don't forget to register yourself, even if there isn't anyone else registered, so that anyone who is newly diagnosed can find you.
bit.ly/rrcontact

Rare Connect

Rare Connect is a free online platform with condition specific communities and general discussion groups.

The platform was set up by EURORDIS: an alliance of rare disease patients in Europe. Discussions are translated across multiple languages.
bit.ly/rrrareconnect



Look for the information standards logo.

The information standard is a programme set up to regulate health and care information. If an organisation is displaying the information standards logo, you know you can trust the information.

Specialist in the condition.

When trying to find information on a really rare condition, it may be helpful to find a specialist in the condition. Experts often write articles for journals about conditions, detailing cases they have seen, ways they have treated symptoms, etc. You may want to look for authors of papers on the condition, as they will be experts.

Know all the names that people use for a condition.

You can do this by going to Orphanet and searching for your condition. Orphanet is a European website providing information about rare diseases. When you have found the right page you should see that in the blue table there is a list of other names for the condition.
bit.ly/rrorphanet

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