THE BRITISH PORPHYRIA ASSOCIATION
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GENETIC ALLIANCE UK

Seeking A Rare Diagnosis | Children

This guide is for people who think their child may have a rare condition and are seeking a diagnosis for them using NHS services in England.

In this information we have used the term 'your child' but we recognise that there are a variety of relationships (e.g. foster child,

child cared for by a guardian/grandparent, etc.) This guide gives information on seeking a rare diagnosis for a child.

If there are words or terms in this information that you don't understand, please use our Genetic Alliance UK's glossary which explains genetic and medical terms in plain English.

Genetic testing for children | what to expect

Prepare for your child's genetics service 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 will often ask you to bring your child with you to the appointment if you are seeking a diagnosis for them.

The history of the symptoms your child currently has or has had in the past is a key part of the assessment. You may want to think about this before your appointment and make a list. Focusing on these things can sometimes feel upsetting, but it can build a picture of your child's symptoms.

You may also want to make a list of any medication that your child is taking or has taken in the past to share at your genetics service appointment.

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 friend for support. They may also remember points about the appointment that you miss.

It's OK to ask for an interpreter if you need one. It is important that this is arranged before the appointment.

Top tips

- Before attending appointments, it is a good idea to write down any questions that you want to ask when you are there. It can be helpful to take a notebook with you.

- Some people record the appointment on their phone so that they can listen again afterwards. You should 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 healthcare professional to write them down for you to look at again later.

- You can also ask if you can have access to your child's electronic record as this is now available at many hospitals.

At your child's genetics service appointment

Often at the start of the first appointment, you will be asked what you are hoping to find out, for example:

- A diagnosis for your child
- How your child might 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 collecting information to help with the 'genetic assessment process'. This is a diagnostic process where clinical geneticists (doctors who specialise in diagnosing and managing genetic conditions in individuals and families) work with laboratory staff and other medical professionals to explore the possible reasons for your child's symptoms.

There are several different types of possible genetic changes. The cause of the condition might be a change in a single gene, or it might be that a chromosome has a piece missing (deletion) or an extra piece (duplication), or it may be that pieces of chromosomes have 'swapped around' (translocation).

Finding the genetic cause of your child's symptoms can happen through a combination of investigations:

- Asking about your child's birth and development in detail
- Asking about your child's family medical history
- Physical examination potentially including photographs
- Laboratory investigations (if necessary, blood samples are taken during the appointment or shortly afterwards)
- Taking measurements, doing scans, etc.

Asking about your child's development in detail

The history of your child's development and the pattern of symptoms that they currently have or have had in the past is a key component of the assessment. You may want to think about this before your appointment and make a list. Focusing on these things can sometimes feel upsetting, but it can build a picture of your child's symptoms.

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. If you have your child's 'red book' which captures your child's key development milestones and growth pattern then take it with you to the appointment.

Asking about your child's family medical history

It is likely you will be asked about your child's family history to see if patterns can be found that might give clues to the cause of their symptoms.

You might find it difficult to describe your family history if your child is adopted or you have little contact with extended family members.

Family history is important because if the condition is genetic, then it may be the result of changes in genes or chromosomes that were passed down from one or both parents. However, many conditions can occur for the first time in a child, even when they are genetic. This is called 'de novo'.

Sometimes it may be possible to make a diagnosis based on these historical descriptions alone without the need for genetic testing.

Physical examination potentially including photographs

The aim of a physical examination is to identify any physical features that might give a clue about a possible diagnosis. Photographs may be taken for your child's medical records and can be useful for monitoring changes over time.

The healthcare professional performing the examination should explain why the examination is needed and take steps to make sure you and your child feel comfortable throughout the process. You may also be asked to sign a consent form.

Laboratory investigations

Once the healthcare professional has an idea about a potential diagnosis for your child, they may order laboratory tests straight away to try and confirm the suspected diagnosis.

Samples of your child's DNA (and sometimes the parents' DNA) will be taken to test and look for changes. The samples needed can vary and may include blood, saliva or skin. Other tests may include a scan of the kidneys or heart, x-rays or a specialist eye test.

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

After the appointment

The genetics professional you see will usually write to you after the appointment summarising what was discussed and what is planned so that you have a written record of the appointment.

There can often be a long wait for results. Learn more about genetic testing by visiting the 'genetic services and testing' section of our website.

Rareminds Wellbeing Hub has a ‘Navigating Healthcare’ section with guidance on appointments and waiting.

Additional information

You may find it helpful to read this guide from Unique that explains what happens during a clinical genetics/genetic counselling appointment. Unique are a charity who provide support, information and networking to families affected by rare chromosome and gene disorders.

What is genetic counselling?

You may be offered genetic counselling as part of your genetic testing. Genetic counselling is a service that provides support and information on genetic conditions.

Genetic counsellors are highly trained healthcare professionals who are skilled at helping you think through what having a genetic test or being diagnosed with a genetic condition might mean for you and your family.

It may involve:

- Learning about a health condition that runs in your family, how it is inherited and how relatives may be affected.

- An assessment of the chance of your child having an inherited condition.

- A review of the medical history of your family or your partner's family and drawing up a family tree.

- Support and information if you have a child affected by an inherited condition and you want to have another child.

- Going through options that might be available to you in terms of testing a future pregnancy (also referred to a prenatal diagnosis). This is a very personal choice and you will be fully supported in whatever decision you make.

- 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.

- Being given clear, accurate information so you can decide what are the best options for you.

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

Genetic tests are complex and it can take a long time to get the results back from genetic testing.

It is worth asking whether there are any relevant research studies carrying out genetic testing, but there is no guarantee that this would return results more quickly.

Your genetics professional or another specialist may be able to update you on research trials that are already underway or are planned for the future.

There are also several websites with information on research into rare conditions:

- National Disease Registration Service
- Be part of research

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 called ‘watchful waiting’. It can be frustrating and worrying waiting for answers. It can be helpful to ask your healthcare professional to explain how and when they will provide you with updates.

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 a dedicated support community available for families of children and young adults with undiagnosed genetic conditions in the UK.

Rareminds Wellbeing Hub has a ‘Navigating Healthcare’ section with guidance on appointments and waiting.

Your healthcare rights

Sometimes people report that they are not happy with the support they have received on their journey to diagnosis. This might be because people don’t feel they have been listened to, or their wishes have been ignored. Sometimes it is because important steps in the process have not been clearly explained or communicated.

In England, the NHS Constitution sets out the rights you have as a patient of NHS services. You have the right to be involved in decisions that affect you and NHS staff should treat you with

kindness, dignity and respect. You have the right to complain if things don't go as you expect.

For information on how patients can give feedback or make a complaint about NHS care or treatment, see the NHS website for guidance on how to complain to the NHS. It includes information on the complaints arrangements, and what to expect when making a complaint.