THE BRITISH PORPHYRIA ASSOCIATION
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GENETIC ALLIANCE UK

Seeking A Rare Diagnosis | Adults

This guide is for adults seeking a diagnosis of a rare condition using NHS services in England.

Seeking A Rare Diagnosis resources created by Genetic Alliance UK with funding from Novartis

In this information we have used the term ‘you’ but we recognise that there are a variety of relationships (e.g. you could be seeking a diagnosis for yourself, you may be a carer for an adult who is seeking a diagnosis of a rare condition, etc). This guide gives information on seeking a rare diagnosis for an adult.

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 adults | what to expect

It is important to consider the range of genetic services and tests that might be available in your situation.

There are lots of reasons why someone might get a genetic test, including:

- Your doctors think you might have a genetic condition and they want to do a genetic test to find out.
- People in your family have a specific genetic condition which might mean that you also have it.
- You are pregnant and a genetic test could reveal if the baby has a genetic condition.

Types of genetic test

There are different types of genetic tests available through the NHS. These include:

- Diagnostic genetic tests are for when doctors think that someone might have a genetic condition based on symptoms that they have. Depending on the symptoms or circumstances, the test might check for a single specific genetic condition or for many different conditions.

- 'Carrier' genetic tests find out if someone carries a genetic change but might not have the condition themselves. These conditions are called 'recessive'. If a person carries the genetic change, there is a chance that they will pass it onto any children that they have but the child will only usually have the condition if the genetic change is passed to them by both the mother and father.

- Predictive genetic tests tell us whether someone is likely to develop a specific genetic condition in the future even when they have no symptoms when they are tested. This is usually done when someone else in the family has already been diagnosed.

- Pre-implantation genetic testing (PGT) is something that can be done as part of an in vitro fertilisation (IVF) procedure. It is used to help people who have a high likelihood of passing on a severe genetic condition to their child.

Your doctor will discuss which type of genetic test is most appropriate with you.

You can read more about the benefits and disadvantages of genetic testing in Genetic Alliance UK's guide to genetic services and testing.

Prepare for the 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.

Before your appointment, it can be helpful to think about the history of the symptoms you currently have or have had in the past. Listing your symptoms and how they affect you can help your doctor build a picture of your symptoms.

It may also help to have a list of any medications you are taking and have taken in the past.

You may wish to take along a family member or friend for support at the appointment. They may help you to remember parts of the appointment that you miss.

It's OK to ask for an interpreter if you need one. It is important to arrange this 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, so it can be useful to take a notebook with you.

- You might want to record appointments on your phone so that you can listen again afterwards. You should ask permission from the other people at the appointments before recording them.

- 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.

At the 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

- How you might be affected in the future

- If any current or future children could be affected

- Whether other members of your 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) work with laboratory staff and other medical professionals to explore the possible reasons for your 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 symptoms can happen through a combination of investigations:

- Asking about your symptoms
- Asking about your 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 symptoms in detail

The history of the symptoms you currently have or have 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 symptoms.

It can be frustrating having to repeat your medical history to yet another professional, but this is a very important part of the assessment.

Asking about your family medical history

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

You might find it difficult to describe your family history if you were 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 with your consent for your health record.

The healthcare professional performing the examination should explain why the examination is needed and take steps to make sure you feel comfortable throughout the process.

Laboratory investigations

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

Samples of your 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 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 the Genetic Alliance UK website.

'I felt I had been going around in circles with symptoms that were not being believed or understood for years, however once I saw the right person everything went smoothly in relation to diagnosis and genetic testing.'
Participant, Rare Experience 2020 Survey

Top tips

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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(ren) having an inherited condition.

- A review of the medical history of your family and drawing up a family tree.

- If relevant, going through options that might be available to you in terms of testing a future pregnancy (also referred to as 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.

Sometimes it is not possible to undertake genetic testing straight away.

In these situations, you will usually be invited back for follow-up appointments several years later when there could be more information because your condition may have developed further, 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.

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.