

Seeking A Rare Diagnosis | Adults

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

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 [Genetic Alliance UK’s glossary which explains genetic and medical terms in plain English](#).

How do I get a diagnosis?

If you have health concerns about yourself or someone you care for, it is a good idea to discuss them with your GP.

Your GP should listen and then discuss your concerns with you. They may do an examination and do some tests, such as taking blood or checking blood pressure. If they are unable to offer a diagnosis, they may refer you to other specialist healthcare professionals for their opinion.

If a genetic condition is suspected by your GP 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.

Your GP may also refer you to a nurse or a healthcare professional who can help with symptoms. For example, if there is muscle weakness then you may be referred to a physiotherapist. A physiotherapist is a healthcare professional who helps people using movement, exercise, massage, advice and other techniques. Nurses and healthcare professionals may be able to help you obtain a diagnosis.

Genetic services offer a range of care and cover agreed areas nationally. [More information on Regional Genetic Services is on Genetic Alliance UK's website.](#)

In some cases, it might take some time to find a medical diagnosis. This can be due to symptoms being common in a number of different conditions, making it difficult to know which condition you may have.

Prepare for the GP appointment

- Before the appointment, it is a good idea to think about what your GP needs to know. They will want to know about symptoms, when they started and how often they happen. It is also helpful to explain how the symptoms feel and how they are impacting your life. It is important that you tell your GP about all the symptoms, even if you may find them embarrassing to talk about.
- Some people with rare conditions find it helpful to record symptoms in a diary. There are also apps available for smartphones which can help you track symptoms.
- If any family members have had similar symptoms and experiences, or if there are conditions that are common in your family (such as heart disease or muscle weakness conditions), you may want to tell your GP about them.
- If you suspect that you may have a particular rare condition, tell your GP.
- Tell your GP if a family member has already been diagnosed with a known genetic condition and if genetic changes were identified by specialist testing.
- Remember to tell your GP about any other existing health conditions and tell them about any medicines currently taken or that have been taken in the past.

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

At the GP appointment

- During the appointment, if you don't understand something then ask your GP to explain or write it down for you. It's OK to ask your GP to repeat something you haven't heard or understood fully.
- You might want to record the appointment on your phone so that you can listen again afterwards. You should ask permission from the other people at the appointment before recording it.
- If you feel you need support during the appointment, you can take a friend or family member with you.
- At the end of the appointment make sure you are clear about what the next steps are, what you are expected to do (for example, you might need to phone for test results) and what your GP is expected to do. You can ask your GP to write down a summary of what will happen next.
- Hopefully, you will feel that your GP has listened to you and is taking action to find out what is causing the symptoms. However, if you feel that your GP hasn't listened to you or has ignored the information you have given them, you can ask for another opinion. [The Patients Association has a helpful guide on seeking a second opinion.](#)

- Remember, it can take a long time to receive a diagnosis. The ‘diagnostic odyssey’ is a term used to describe the time between first symptoms and receiving a final diagnosis. This time can be challenging and worrying, but there is support available. [Rareminds Wellbeing Hub](#) has sections on diagnosis, uncertainty, difficult feelings, navigating healthcare and sources of support.

How to access NHS genetic services and testing for adults

There are genetic services and tests available through the NHS.

To get a genetic test on the NHS, your doctor must think it is likely that you have a genetic condition or that you could pass a genetic condition on to your child. A doctor might ask for information about things like family history of genetic conditions or any symptoms that you have.

If you think you might have a genetic condition or that you might pass a condition onto your child, it is important to speak to doctors

so that you can be assessed for NHS testing. This ensures that the most appropriate genetic test is undertaken and the results are explained to you by the doctor who arranges the test.

If you are referred to NHS genetic services, you might speak with a genetic counsellor. These are skilled professionals who you can talk to about genetic condition diagnosis, such as the risks and benefits

of a genetic test and exactly what it would mean for you and your family to get a diagnosis or not.

Genetic services offer a range of services and cover agreed areas nationally. Genetic Alliance UK's website has [information on Regional Genetic Services](#) as well as [information on direct-to-consumer genetic testing](#).

Depending on your situation, the first step to getting a genetic test on the NHS is usually to speak with your GP in order to get a referral to genetic services. If you have family members who are already diagnosed with a condition, it might be that you can speak to genetic services directly.

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.