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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 Genetic Alliance UK's glossary which explains genetic and medical terms in plain English.

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

How do I get a diagnosis for my child?

If you have concerns about your child’s health you can talk about them with your GP or your health visitor.

Your GP or health visitor 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 healthcare professionals.

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.

Your GP may also refer your child to a nurse or a healthcare professional who can help with symptoms. For example, if your child is experiencing muscle weakness then they 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 for your child.

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

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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.

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

Top Tip

On your journey to diagnosis, you may find that you have to wait a long time for an appointment or test. You may be placed on a waiting list. This can be frustrating and you may be unsure how to seek help while you are waiting. NHS England has launched the My Planned Care website which provides information about waiting times and where you can access support while you wait.

Prepare for your child's GP appointment

- It is a good idea to think about what your GP needs to know before your appointment. They will want to know about your child's symptoms, when they started and how often they have them.

- Take along any record of dates of when your child met development milestones such as sitting, smiling, walking, talking, etc. 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.

- If any family members have had similar symptoms and experiences or if there are conditions that are common in your child’s family, you may want to tell your GP about them.

- If you suspect that your child may have a particular rare condition, tell your GP.

- Remember to tell your GP if your child has any other existing health conditions.

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

- Remember to tell your GP about any medication that your child is taking or has taken in the past.

At your child’s 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 take some notes during the appointment, or maybe even record the discussion on your phone to review later. 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 making your child unwell. 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.

How to access NHS genetic services and testing for children

There are genetic services and tests available through the NHS. You might be offered NHS genetic services if your doctors think that you or your child might have a genetic condition or think that you might pass a condition onto your child.

To get a genetic test on the NHS, your doctor must think it is likely that you or your child has 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 your child has.

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 your child is referred to NHS genetic services, you might speak with a genetic counsellor. These are healthcare 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.