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.

What can getting a diagnosis mean for me, or my child?

A diagnosis can be important on a practical and emotional level. Having a genetic diagnosis can help you to:

- Understand your child's condition and have a sense of how the condition may affect your child in the future
- Identify any health issues that need to be monitored
- Make informed care decisions, including identifying appropriate services, potential treatments and relevant clinical trials
- Understand whether others in your family will be affected by the condition and help with family planning decisions
- Access services and support for your child's needs, for example educational support or social care services.

- Find support organisations to help provide information, support and reduce feelings of isolation

- Explain to other people in your life about your child's health condition.

However, a diagnosis does not always mean things will change. You may find that your child's care plan remains the same and you may continue to be supported by the same healthcare professionals.

Contact is an organisation for families with disabled children. Contact's website has information and support on lots of subjects including education and learning, social care and family life, work and childcare.

Unique are a charity who provide support, information and networking to families affected by rare chromosome and gene disorders. Unique's guide called After diagnosis: what happens next? offers information and support following diagnosis of a genetic or chromosome condition for your child.

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

SWAN UK community member

If your child has received a diagnosis of a rare condition, you may feel disappointed with the information you receive. For many rare conditions, information about the condition and its prognosis (the likely course of a medical condition) may be limited and you may find that you still have unanswered questions.

‘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 community member

Having a diagnosis can help healthcare professionals better understand your child’s condition and help them find the information they need to support you.

‘I was relieved that all of our child’s seemingly unconnected symptoms could be explained by one diagnosis. My biggest fear the entire time had been that we would never find out what was wrong with her.’

Parent of child with rare genetic condition

It is important to acknowledge that not everyone will receive a diagnosis. Some people may not be able to access the tests necessary to identify their child’s condition, while others may be affected by a condition that is so rare it is yet to be understood by science. Without a diagnosis, you should still be able to access services and support based on your child’s needs. For example, social care services and education support are intended to be provided based on a person’s needs, not their diagnosis. Contact’s website has information on accessing on social care.

Receiving a diagnosis of a rare genetic condition will never be easy. Discovering that your child has a health condition that might be life changing or life limiting is likely to be an emotionally challenging experience.

Rareminds Wellbeing Hub has sections on diagnosis, uncertainty, difficult feelings, navigating healthcare and sources of support.

'It was an emotional time with lots of suggested conditions that were tested for (which were negative) before finding the right diagnosis. Some doctors acted as if we should be elated at receiving a diagnosis not recognising the enormity and devastation of having your baby diagnosed with lifelong multiple complex health problems. It wasn't a relief to get a diagnosis, it was devastating.'

Participant, Rare Experience 2020 Survey

Why are some conditions difficult to diagnose?

Genetic conditions can be difficult to diagnose. There are three main reasons for this:

- It is the 'rarest of the rare' - a condition that hasn't been seen before and therefore there is not a test for it.

- It is an unusual presentation of a known condition – your child's symptoms might be different to those of other people 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 someone's DNA or genetic make-up but these might not be the cause of their health concerns. The meaning of the result is unclear.

Trying to get a diagnosis for your child can be a worrying time. There are people in your life, healthcare 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. Your child may be referred to a specialist nurse or genetic counsellor who can work directly with you and your family to offer genetic information and support you to make decisions.

There are also patient groups and support organisations that can provide emotional and practical support for you and your family. On our website you can find an A-Z list of patient support organisations

Coming to terms with a diagnosis of a genetic or rare condition, or with not finding a diagnosis

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 or rare 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 people 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 support groups for advice on the help and counselling they may be able to offer.

If your child has brothers or sisters, they may feel worried about their sibling. The organisation Sibs provides resources and support for people who grow up with or have grown up with a disabled brother or sister.

The journey of seeking a rare diagnosis and coming to terms with any outcomes can potentially be stressful. We all handle stress differently and you can contact your GP if you feel stressed or anxious - they will help with getting support.

Another potential source of support is Rareminds Wellbeing Hub which has sections on diagnosis, uncertainty, difficult feelings, navigating healthcare and sources of support.

Unique also has a range of practical guides, as well as genetic condition specific guides. Even if they don't have a relevant guide, you can join them and request further information. You can also join their private Facebook café to interact with families who have had or are having similar experiences.

Top tips

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 a support group

For some conditions there may be a support group available. Support groups can provide information about a condition. Support groups often have information on their websites and could put you in touch with parents of a child with the same diagnosis.

Genetic Alliance UK's website has information on support groups in their alliance. If you can't find a relevant organisation then please contact Genetic Alliance UK.

Unique can also connect you with sources of support.

Additionally, there may be local organisations and support services that can help you. You can enter your postcode on the GOV.UK website to find services in your local community.

Online forums

The internet is home to lots of virtual areas for people to meet and talk about their experiences with genetic, rare and undiagnosed conditions.

Be aware that content in online forums and social media may be opinion rather than factual. Ensure you check the quality and suitability of what you read in online forums and on social media.

As a free, informal platform Facebook is home to a number of online support groups. It can be comforting and helpful to talk to someone with, or who has a child with, the same condition – someone who has had similar experiences.

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 Facebook to join and talk to people. If there isn't already a Facebook group for your child's condition, you can always set one up for the next person who is searching for one. It can be a great way to connect with others directly.

Research

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

Speak to other people

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

Additional information

Rareminds Wellbeing Hub has sections on diagnosis, uncertainty, difficult feelings, navigating healthcare and sources of support.

The NHS Mental Health website has lots of guidance and support for your mental health.

Mind's Information and Support website hub has lots of advice and also shares details of their helplines.

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.