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.

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.

What can getting a diagnosis mean for me?

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

- Understand your condition and have a sense of how the condition may affect you or your family 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 needs, for example workplace 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 health condition

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

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

SWAN UK community member

If you have 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… what I got was a string of letters and numbers.’

SWAN UK community member

Having a diagnosis can help healthcare professionals better understand your condition. It can also help healthcare professionals to find the information they need to support you.

‘Getting my diagnosis was in many ways reassuring as at least I knew what I was actually having to deal with and how I could start to adjust to life with my rare condition.’

Patient and Public Involvement representative

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 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 needs. For example, social care services and workplace support are intended to be provided based on a person’s needs, not their diagnosis. For example, social care services and workplace support are intended to be provided based on a person’s needs, not their diagnosis. Information on getting a needs assessment can be found on the NHS website.

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

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 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 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. You 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 you have a health condition can be difficult.

The news that you have 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

‘An undiagnosed label helps as a starting point - it’s difficult when you don’t know where you fit in to know what’s right for you.’

Participant, Rare Experience 2020 Survey

Every person deals with diagnosis, or the news that they may remain undiagnosed, differently. There is no right or wrong way.

‘Finally getting a diagnosis was a huge help to my family and friends as they could then start to understand what to expect from me.’

Patient and Public Involvement representative

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 specific needs.

‘Looking back, I guess I went into a state of grief when we got a diagnosis.’

Workshop participant

‘Reflecting on my diagnosis, initially I went into a state of disbelief, then a period of mourning the person I once was or aspired to be. Finally I achieved acceptance and started to be the new version of me.’

Patient and Public Involvement representative

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.

‘Even if you receive a diagnosis, it won’t necessarily bring any answers. Once we finally received the diagnosis, we were plunged into a world of unknowns. It was a super rare disease and there was absolutely nothing in medical literature about the condition. We felt utterly alone. It was only when we managed to find a tiny Facebook group with other diagnosed families that we found a community. That was our lifeline in a sea of silence.’

Genetic Alliance UK member representative

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.

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

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.

These services offer confidential support from trained volunteers. You can talk about anything that's troubling you, no matter how difficult:

Call 116 123 to talk to Samaritans, or email: jo@samaritans.org for a reply within 24 hours

Text "SHOUT" to 85258 to contact the Shout Crisis Text Line, or text "YM" if you're under 19

If you're under 19, you can also call 0800 1111 to talk to Childline. The number will not appear on your phone bill.

These services will only share your information if they are very worried about you or think you are in immediate danger.

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 people who have 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 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 people who have had or are having similar experiences.

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. It's also important to remember that information on online forums may not be accurate, or relevant, to people living in the UK. 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 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 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 can be reassuring and helpful. Support organisations can help to connect you with other people.

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.

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 people who have had or are having similar experiences.

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.