

Diagnosis of rare conditions

The 'diagnostic odyssey'

Many people with a rare condition face challenges in getting a diagnosis, with more than a third of people having to wait more than five years for an accurate diagnosis. This 'diagnostic odyssey', the time between first symptoms and receiving a final diagnosis, can have a significant impact on a person's quality of life and wellbeing.



How a person is supported on their journey to diagnosis is important. Too often, people with rare conditions will feel unsatisfied with the level of information and support that they are provided.

Understanding the experience of diagnosis

Genetic Alliance UK published [Good Diagnosis: Improving the experiences of diagnosis for people living with rare conditions](#) in February 2022. The report followed a series of workshops with people with rare conditions designed to understand their experience of diagnosis and to identify the key principles of a 'good' diagnosis experience.

Central to a good diagnosis experience are healthcare professionals. No healthcare professional can be aware of every rare condition, but ignorance of rare conditions generally leads to **slower referral**, **slower diagnosis**, **misdiagnoses** and **slower access to appropriate specialised care**.

Principles of good diagnosis

There is no one right way to pursue or receive a diagnosis, what works for one person may not be appropriate for another. However, the Good Diagnosis report identified that the diagnosis experience benefits from 8 principles. The next page outlines these further.



A diagnosis that is timely and accurate



Delays in diagnosis and receiving misdiagnosis significantly affect a person's mental and emotional wellbeing. A fast and accurate diagnosis can mean greater treatment choice and support informed decision-making, leading to people with rare conditions being able to better manage their condition. Shortening the journey to diagnosis and avoiding often costly unnecessary referrals, tests and treatments will benefit NHS Services.



A diagnosis that is coordinated and collaborative



People want to feel that they are an active and valued partner in their care – working with their healthcare professionals in a coordinated manner to ensure the person's health, information and emotional needs are met. A diagnosis care plan could be a helpful tool in ensuring individuals are informed, consulted and able to participate in the decision making around their diagnosis.



A diagnosis that is informed and supported



The experience of diagnosis can be improved by ensuring people have access to reliable sources of information. It is important that healthcare professionals know how and where to signpost people living with rare conditions. Support organisations play an integral role in providing high quality support and information to people throughout their diagnostic journey.



A diagnosis that is acknowledged and respected

It is common for people with rare conditions to continue to experience challenges after they receive a diagnosis. This often comes from healthcare professionals having little experience or understanding of their condition. This can result in delays in being referred to appropriate specialists or services, delays in getting access to treatments and can contribute to a breakdown in trust between the person and those responsible for delivering their care.

