**What is the diagnostic odyssey?**

**Genomics 101 explainer episode transcript**

# **Florence: What does it mean to go on a diagnostic odyssey? I'm joined by John Pullinger, Senior Bio Sample Operations Manager for Genomics England to find out more. So, John, first of all, can you explain what we mean by diagnostic odyssey?**

**John:** Yes, of course. The diagnostic odyssey is a term used to describe the journey that many people with rare conditions and their families undertake to receive an accurate diagnosis, a journey that takes on average over five and a half years.

The rarity of the condition means that there are few, if any, other people affected by it, for doctors to draw their experience from. Some individuals might never receive a diagnosis.

My job involves making sure that samples sent through the Genomics England processes can travel smoothly from the NHS hospitals to be sequenced and the results be reported back to the individual. We try and minimise the amount of time that samples and associated data is in our care.

**Florence: And for people listening who might not know, could you explain why it sometimes takes a long time for people to receive a diagnosis?**

**John:** There are estimated to be over 7,000 rare conditions.

This means that healthcare professionals may not be familiar with all of them and so may not recognise them or know how to test for them. In addition to this, some conditions affect multiple parts of the body. For example, skin, the heart, and the lungs. In these cases, there will be a need to visit specialists from multiple departments, and each will be looking specifically at their own area.

This could lead to referral loops where the patient needs to consult multiple healthcare professionals, all of which contributes to the time taken to receive a diagnosis. Since, for the majority of rare conditions, there is an underlying genetic cause. This means that most individuals who get a diagnosis will receive one through genomic testing, whether that be whole genome sequencing as offered here at Genomics England, or more targeted panel testing.

Typically testing will identify a particular gene, which is known to be linked to a specific condition. For certain conditions, it requires a real expert in the condition to even think about testing for it. Sometimes a condition will present in a way that is different to most other people who have it. So they may have symptoms that others don't. This also adds to the buildup of time taken to receive the diagnosis.

**Florence: So, you mentioned earlier, John, that the diagnostic odyssey lasts an average of five and a half years. Can you explain what kind of effect this long waiting time has on individuals and their families?**

**John:** Absolutely. One aspect of the diagnostic odyssey that is important to recognise is the physical effect of the as yet undiagnosed condition that's present and affecting the individual and their family on a daily basis. Those with rare conditions may be affected by a range of emotions connected to the ongoing journey that they're on, including feelings of isolation.

Also stress and anxiety. The fear of unknown can have a massive knock-on effect on the mental health of the individual and their family. And it's important to recognise the signs of this so that people can take steps to manage their mental health. Many rare conditions first present themselves in children and young adults, so considering the effects on their day-to-day lives is especially important.

**Florence: If you'd like to learn more about how the diagnostic odyssey can affect someone, listen to our previous podcast, “Hope for those with no primary findings”, where Participant Panel member Lisa Beaton, shares her experience of awaiting a diagnosis for her daughter. And so, John, can we talk now about what happens at the end of a diagnostic odyssey?**

**John:** A section of the odyssey that is essential to understand is potentially getting a diagnosis. It may come as a surprise to think that the diagnosis can sometimes be scary as well as a potential relief to the family and also the individual involved. But this reason the work of genetic counsellors is crucial to help those with rare conditions, understand and adapt to the medical, psychological, and potential reproductive implications of their new diagnosis.

**Florence: Our previous podcast, “The impact of a genetic diagnosis on mental health” covers this topic in much more detail. So for my final question today, I wanted to ask whether there are ways that families or individuals affected by rare conditions can access support.**

**John:** We would recommend that anyone who might be going through a diagnostic odyssey who wants to know more about their care to contact their doctor or other healthcare professionals in their genetics team, additional resources are also available online, including the NHS website and charities such as Genetic Alliance UK and SWAN UK.

There are also lots of brilliant patient communities and groups that you can get support from.

**Florence: That was John Pullinger explaining what it means to go on a diagnostic odyssey. If you'd like to hear more explainer episodes like this, you can find them on our website at** **www.genomicsengland.co.uk****.**

**Thank you for listening.**