

Experience of diagnosis in mitochondrial disease

Plain Language Summary

Why we did this research

Mitochondrial diseases, or 'mito', can cause a wide range of symptoms with different levels of severity. This can make getting a diagnosis a challenging and lengthy process for people living with mito, and their families or caregivers.

It is therefore important that people are well supported before, during, and after their diagnosis, to help look after their wellbeing during what can be a difficult time in their lives.

Our research aimed to better understand people's **experiences during the process of getting a diagnosis** of mitochondrial disease. In particular, we were interested in whether the diagnosis was explained to patients, and where relevant, their parents or caregivers, in a clear and supportive way.



What the study involved

Our team reviewed the current published research, which showed that although receiving a diagnosis can have a positive impact overall, many people have to see multiple specialists and can experience long delays and different diagnoses along the way.

To build on these findings, we ran an **online survey** where patients and their relatives or caregivers shared their experiences **before, during, and after getting a mito diagnosis**. The study was conducted in the UK over April/May 2025.



Who took part

A total of **103** people took part in the study.

44 patients

50 parents

6 partners

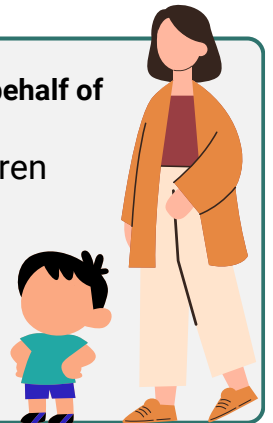
59 caregivers

3 family members

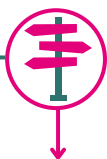
We had **responses from or on behalf of**

55 adults and **27** children

who ranged from 7 months to 77 years old, with 21 responses given on behalf of people who had passed away.



What we found: **Before diagnosis**



A long journey to diagnosis

On average, it took more than 8 years from first having symptoms to getting a mito diagnosis, with participants usually starting this journey because of fatigue, vision problems, or co-ordination difficulties.



Multiple specialists

Most participants saw multiple specialists, with some seeing up to 11 different doctors before getting a diagnosis.



Misdiagnoses

Over a third of participants were misdiagnosed with other conditions such as diabetes, mental health issues, and cerebral palsy.

During diagnosis



Although most of the participants were given their diagnosis in person, around 30% were told over the phone, by a letter, or by having results uploaded to the NHS app.



The number of people who felt confident in their diagnosing doctor's experience with mito has dropped in recent years.

These people were less likely to feel positive about their overall diagnosis experience and were more likely to feel that their diagnosis was given insensitively or with a lack of expertise.



Three quarters of participants felt confident before 2020, compared with less than half of the participants diagnosed in the last 5 years.

A possible reason for this is that genetic testing has become much easier to access. While this is a positive step, it also means that testing can be arranged by doctors who do not specialise in mito, which may impact how confident some people feel about their doctor's level of experience.

After diagnosis



People generally had a positive experience after their diagnosis, feeling able to ask questions and supported by their care team in decisions about their physical health.



However, feeling alone was common, with many getting no information about patient organisations or psychological support options to help process the emotional side of their diagnosis.



Less than 60% of participants received a referral to a specialist clinic. Some of these had to wait up to two years for their first appointment, with little information or psychological support offered during this time.

What this means

This study identified several key areas for improvement in mito diagnosis, which may be contributing to uncertainty and emotional distress for patients and their families.

What needs are not being met?

Many patients felt they were **not being listened to** by their doctors, with their concerns dismissed without investigation or understanding.

When doctors appeared less familiar with mito, **confidence in care** was reduced.

Sensitive delivery of the diagnosis is important. Some patients felt unprepared if a diagnosis was delivered unexpectedly.

Many people felt that **mental health support** was limited.

What can be done?

Feeling heard and taken seriously may help patients feel more supported and could contribute to **faster testing**.

Patients prefer to receive their diagnosis and care plan from an **appropriate specialist** with strong **professional mito awareness**.

Clearer information in advance may help patients feel more prepared for diagnosis conversations.

Improving access to **psychological support** throughout the diagnostic journey could help patients and their families feel better supported.

What happens next?

In the next phase of this research, we will conduct in-depth one-to-one interviews with people who took part in the survey. These interviews will help us better understand their individual experiences with getting a mito diagnosis.

We are looking to learn more about what changes could be made to the diagnosis process and what resources could be developed to help support people before, during, and after diagnosis. With this research, we hope to make a real difference to the overall experience of patients and their families or caregivers.