

Genetic testing FAQs

In April 2023, the UK genetic testing guidelines for MND were changed. These guidelines describe who is eligible to receive a genetic test for MND in the UK.

How have the genetic testing guidelines been change?

The UK guidelines (UK National Genomic Test Directory criteria) were updated in April 2023 to state that there is no limit on age or family history for routine genetic testing. However, we are aware that there are significant delays with accessing genetic counselling, testing and results and the MND Association is considering whether and how we are able to assist with this issue.

Why have the guidelines been changed?

Before the change, genetic testing for MND was usually restricted to people with family members who are also affected by MND, or people whose symptoms start before they are 40 years old. A study published in September 2022 showed that under these guidelines, many people with MND who may have a genetic form of the disease, wouldn't be offered a genetic test, so knowledge of the genetic cause would be missed.

The paper suggested that hundreds of people in the UK, and thousands worldwide, could be being overlooked. This means they wouldn't be offered proper genetic advice or counselling, or access gene-based therapies in the future. This change in guidelines now allows more people living with MND have the option to have genetic testing.

Are there treatments that could help people with a genetic cause of MND now?

There are signs that science is going in the right direction in the hunt for effective treatments of MND, as shown by the results from the Tofersen clinical trial. These results provide increasing confidence that Tofersen is having a beneficial effect in people living with SOD1 MND. The Food and Drug Administration (FDA) has recently approved Tofersen (Qalsody) for the treatment of SOD1 MND in the United States. The European Medicines Agency is now reviewing the data around Tofersen and will decide if the treatment could be approved in the UK and Europe.

Tofersen is a genetic therapy that is designed to specifically target the SOD1 gene and so can only work in those people who have a change in that particular gene (around 2% of all people with MND). Therefore, the treatment is only offered to those people who know they carry a genetic change in the SOD1 gene. The only way to know this is to have genetic testing. This is a very real demonstration of how useful genetic testing could be in the future.

Are there other potential treatments like Tofersen being tested for genetic MND?

There are several gene therapies currently being tested in clinical trials. These clinical trials include experimental treatments for SOD1, C9orf72 and FUS genes, and it is likely that more of these gene therapies will emerge over the coming years. Changing the current guidelines to allow everyone diagnosed with MND to be offered genetic testing, alongside genetic counselling, will allow more people living with MND an opportunity to be involved in clinical trials and ultimately to access genetic treatments once they are available.

How and where can I get tested if I want to?

You will need to speak to your healthcare professional about genetic testing.

It may be possible to arrange for genetic testing through a private clinic. You will need to speak to your GP about a possible referral.

What are the pros and cons of having genetic testing?

Genetic testing has many implications and should only be undertaken with careful consideration, appropriate genetic counselling and an understanding of the potential consequences.

Your healthcare professionals will be able to discuss some of this with you and genetic counsellors will be involved in any decision you may make about whether to have genetic testing.

Do I have to have genetic counselling?

It is imperative that testing is supported by genetic counselling. Specially trained counsellors can offer tailored support, understanding and explanation at each stage of the process so you are able to make informed decisions.

I already have MND. Can I be genetically tested?

You will need to speak to your healthcare professional. Neurologists may be able to arrange testing on a case-by-case basis. You may also be able to arrange for genetic testing through a private clinic. You will need to speak to your GP to discuss a referral. Any genetic testing should be carried out in conjunction with genetic counselling.

Will the Association be campaigning for genetic testing to be more widely available?

We engage with the government and other key stakeholders such as the NHS on a wide range of issues and are continually reviewing our priorities. In light of these new findings, we will consider how best to raise the issue of access to genetic testing as part of our planning processes.

Where can I find more information?

[Living with motor neurone disease \(MND\) guide \(mndassociation.org\)](https://www.mndassociation.org)

Page 10-12 covers genetic counselling and testing

Is the MND Association doing any further work on genetic testing?

We are funding a healthcare research project on the development of a [decision-making aid](#) for people with MND and their families.

Further publications from the MND Association

[B1-Introduction-to-inherited-MND.pdf \(mndassociation.org\)](#)

[B2-Genetic-testing-and-insurance.pdf \(mndassociation.org\)](#)

[B3-Options-when-starting-a-family.pdf \(mndassociation.org\)](#)

Research blog

[Revisited: Should routine genetic testing be considered for all cases of MND? - MND](#)

[Research Blog](#)

[What does the FDA approval of Tofersen mean for the UK MND community? - MND](#)

[Research Blog](#)

Video on genetic testing from the International Alliance

[Genetics – Counselling & Testing - YouTube](#)

[Developing a patient decision aid to support genetic testing in MND | MND Association](#)

[Inherited motor neurone disease \(MND\) - Genetic testing after a diagnosis of MND \(healthtalk.org\)](#)

Statement from Dr Nick Cole, Head of Research, MND Association: “We welcome the changes in the guidelines around genetic testing in the UK National Genomic Test Directory to reflect the removal of age and family history criteria. However, we are aware there are significant delays for people in accessing genetic testing. We are working to consider how we can help remove barriers to testing to ensure more people are able to access genetic testing and appropriate counselling should they wish to.”

MND Association

April 2023