motor neurone disease association

Research

Inherited MND: Genetic testing and insurance

Information Sheet B2

A small proportion (5-10%) of people with MND have a family history of the disease. This form of MND is known as familial, or inherited, MND. Inherited MND is caused by a mistake in the genetic code that holds the instructions for making every protein in our bodies; this mistake may be passed down from one generation to the next.

This publication gives information about genetic testing and insurance for those who have the rare inherited form of motor neurone disease (MND).

This information sheet is **part two** of our information on inherited MND.

- **Part one** of this information sheet looks at how motor neurone disease can be inherited and the importance of taking a full family history.
- **Part three** explains the options available to affected family members who may wish to start a family.

The content is split into the following sections:

Disclaimer: *Please note that information provided in this information sheet is based on a review of the currently available literature. This information sheet was written by the MND Association staff who are not clinicians and so any information provided in this sheet should not be considered as clinical advice. You should always discuss potential treatments with your clinician.*

This symbol is used to highlight **our other publications**. To find out how to access these, see *Further information* at the end of this sheet.

1: Introduction

If you have MND and your GP or neurologist suspects a family history of MND you may want to know which gene is responsible for causing MND in your family by choosing to have a genetic test. This will only allow you to know if you have one of the disease-causing genes which have been identified through research.

If an MND-causing gene has been identified, your unaffected family members may wish to be tested to see if they too have the gene responsible for causing MND in your family. This is known as pre-symptomatic testing.

A diagnosis or family history of inherited MND may affect life and travel insurance, which may be something that worries you and your family. It is important to know that there is a great deal of information and support available regarding this topic.

2: Inherited MND and genetic testing

The gene that is faulty in inherited MND can differ between one affected family and another. Mistakes in genes called SOD1, TDP-43, FUS and C9ORF72 collectively account for about 65 – 70% of cases of inherited MND. Scientists have yet to identify the gene defects that cause the remaining 30%, however, genetic research has been continually growing and making advances over recent years.

If you have MND and the disease is known to run in your family, it is possible to have a routine genetic test to see whether your disease is caused by a mistake in one of the above genes.

I have inherited MND - why might I want to know which gene is causing it?

At the current time, knowing whether or not you have a defect in one of the above genes will not change the progression of your disease or the treatments available to you. Some people might choose to be tested because:

- They have a desire to know exactly what caused the disease. A test result showing that their MND resulted from a particular gene defect can answer the question 'Why me?' once and for all;
- There may be opportunities specifically for people carrying one of these gene mistakes to take part in research studies;
- Much of the current laboratory research into MND is based on the four key disease-causing genes listed above. It is possible that this research may eventually generate a treatment that is more effective for people with these forms of MND. The result of the test has significant implications for the rest of your family.

- If tests show that your inherited MND is caused by any of the mentioned genes, it opens up the possibility of genetic testing for healthy family members to see if they too have inherited the faulty gene. This 'pre-symptomatic testing' should only be undertaken after extensive genetic counselling due to the huge psychological and practical implications (see pre-symptomatic testing below).
- If you do not have a defect in any of the identified MND-causing genes, and you have a family history of MND, then your disease is likely to be caused by a mistake in an unidentified gene. This means that genetic testing will not be available for unaffected family members.
- If there is a family history of MND, and the gene defect causing MND in your family has not yet been identified, it can be very useful for the NHS genetic service to store a DNA sample from affected individuals for future genetic testing. You can ask your GP or neurologist for information regarding this and how it can be arranged. The stored sample may be important for any testing in the future, when more genes that cause inherited MND are found.

3: Pre-symptomatic testing

I am unaffected but I have a family history of MND - can I get tested?

If you are currently healthy but have a relative whose inherited MND is known to be caused by a defect in the SOD1, TDP-43, FUS or C9ORF72 genes, it is possible for you to have a genetic test to see if you have also inherited the faulty gene. This is called pre-symptomatic testing. There is a possibility that if the disease-causing gene in your family has not been identified, then pre-symptomatic testing may not be possible.

Genetic testing is only available to those **over the age of 18** and the result has huge implications. You may find that you do not carry the faulty gene and the threat of disease will no longer hang over you. However, it is equally possible that you find out that you do carry an MND-causing gene.

If you take a pre-symptomatic test it is extremely important that you are then prepared for the possibility of the latter outcome, as the psychological consequences are substantial. For this reason, if you want to be tested, you must be referred for genetic counselling by your GP or neurologist. This will involve at least three sessions with a clinical geneticist before blood is taken for testing. Further counselling should be provided after the test result is given to you (this takes around four weeks from when the blood was taken).

Genetic counselling, although sensitive to the emotional aspects of the situation, is not a form of psychotherapy. A genetic counsellor will explain the facts as clearly as possible, and give you accurate information on the implications for your family. This includes drawing up a family tree, providing information, and discussing any questions

or concerns you have about whether or not you have the disease, or if you are at risk of passing the disorder on to your children.

If you discover that you carry a disease-causing gene, there are currently no preventative measures that can be taken to delay or prevent disease onset (although it is important to note that not every carrier will go on to develop the disease).

For further information about inherited MND, see: Information sheet B1 – *Introduction to inherited MND.*

Family members who decide not to have genetic testing now can always change their minds in future.

If you are an unaffected family member wishing to be tested for an MND-causing gene, you should consider others in your family for whom the result may have consequences. For example, if you are the grandchild of somebody affected by inherited MND, due to a mutation in the SOD1 gene, and are found to carry the SOD1 gene defect, this shows that your parent (i.e., the son/daughter of the affected person) also carries the gene defect. You may need support and advice to help you deal with this, which will be made available to you by your genetic counsellor.

For further information on genetic testing, see the UK Genetic Testing Network resource (find the link in the 'How do I find out more?' section).

4: Inherited MND and insurance

You may be worried that a family history of MND may affect your insurance. If a specific MND-causing gene has been identified in you, or your family, then this gives you an increased risk of developing MND in the future. This may mean that obtaining life and travel insurance cover could be complicated.

Genetic Alliance UK, an organisation that aims to improve the lives of those affected by genetic conditions, has information on life and travel insurance. Their website contains accurate and clear answers covering an array of topics from genetic testing to family history and from an explanation of the Concordat and Moratorium (a policy that guarantees significant insurance cover to anyone who has had a predictive test) to your privacy and what to do if you are rejected by an insurer.

MND Connect can give information on companies who provide travel insurance for people diagnosed with MND.

It is important to note that the information on the Genetic Alliance website address all genetic disorders and are not specific for MND. For instance, in MND, even if you have an MND-causing gene, you may not ultimately go on to develop the disease – this faulty gene may only increase the risk of you developing MND in the future.

For further information and for information about life and travel insurance, please see the Genetic Alliance UK resource (find the link in the 'How do I find out more?' section).

5: Care and support

The care required by someone with inherited MND is no different to a person who has the non-inherited form of the disease and the MND Association has a range of services available to help. Referral to a neurologist who understands the particular needs of people with inherited MND may be helpful.

I am worried about starting a family
If inherited MND is in your family you may have concerns about starting a family and the risks of passing on the gene defect. However there are a number of options that may be available to you.

6: How do I find out more?

Useful organisations

We do not necessarily endorse any of the following organisations, but have included them to help you begin your search for further information.

The contact details are correct at the time of publishing, but may change between revisions. If you need help to find an organisation, contact the Research Development Team (see *Further information* at the end of this sheet for details).

Genetic Alliance

The Genetic Alliance UK is an organisation that aims to improve the lives of people affected by a genetic condition. They have a number of leaflets and documents under the 'Information Centre' tab on their website, including one on insurance.

Address: 4D Leroy House, 436 Essex Road, London, N1 3QP
Email: contactus@geneticalliance.org.uk
Telephone: 0207 704 3141
Website: **www.geneticalliance.org.uk**
www.geneticalliance.org.uk/information/living-with-a-genetic-condition/travel-insurance

NHS National Genetics Education Centre

The NHS National Genetics Education Centre has information for patients and healthcare professionals about genetics.

Address: St Chad's House, 213 Hagley Road, Birmingham, B16 9RG
Email: enquiries@geneticseducation.nhs.uk
Telephone: 0121 695 2529
Website: **www.geneticseducation.nhs.uk**

British Society for Genetic Medicine

The British Society for Genetic Medicine has a directory of UK regional genetic centres so that you can find your local centre.

Address: Clinical Genetics Unit, Birmingham Women's Hospital, Edgbaston, Birmingham, B15 2TG
Email: bshg@bshg.org.uk
Telephone: 0121 627 2634
Website: **www.bsgm.org.uk**

UK Genetic Testing Network

The UK Genetic Testing Network has an information leaflet about genetic testing in the NHS which you can download or view. You can also find out about a specific disease, or test, as well as which laboratories offer these tests.

Address: c/o South East Commissioning Support Unit, National Specialised Commissioning Team, 15 Marylebone Road, London NW1 5JD
Email: SECSU.UKGTN@nhs.net
Telephone: 0203 350 4999
Website: **www.ukgtn.nhs.uk**

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Further information

You may find these information sheets from the MND Association helpful:

2B – *Kennedy's disease*
2C – *Primary Lateral Sclerosis*
2B – *Progressive Muscular Dystrophy*
9A – *Will the way I think be affected?*

B1 – *Introduction to inherited MND*

E – *Research we fund*

We also provide the following guides:

Living with motor neurone disease – our main guide to help you manage the impact of the disease

Caring and MND: support for you – comprehensive information for unpaid or family carers, who support someone living with MND

Caring and MND: quick guide – the summary version of our information for carers

You can download most of our publications from our website at **www.mndassociation.org/publications** or order in print from the MND Connect helpline, who can provide further information and support.

MND Connect can also help locate external services and providers, and introduce you to our available services, including your local branch, group, Association visitor or regional care development adviser.

Helplines STANDARD

MND Connect
Telephone: 0808 802 6262
Email: mndconnect@mndassociation.org
MND Association, David Niven House, 10-15 Notre Dame Mews, Northampton NN1 2BG

Research Development Team
Telephone: 01604 611 880
Email: research@mndassociation.org

MND Association website and online forum
Website: **www.mndassociation.org**
Online forum: **forum.mndassociation.org** or through the website

We welcome your views

Your feedback is really important to us, as it helps improve our information for the benefit of people living with MND and those who care for them. If you would like to provide feedback on any of our information sheets, you can access an online form at: **www.surveymonkey.co.uk/r/infosheets_research**

You can request a paper version of the form or provide direct feedback by email: research@mndassociation.org.

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