Inherited MND

9 in every 10 cases of MND are sporadic, meaning the person has no family history of the condition or genetic link. However, 1 in every 10 people who develop the condition have inherited an altered gene that is known to increase their risk of developing MND. This 10% of cases can be known as inherited or familial MND.

What is an altered gene?

In humans, genes are like an instruction manual that helps our cells develop, maintain themselves and function properly. If a gene contains a significant change, it can affect the products from the instructions in a way that makes them less efficient or unusable.

If you imagine a gene as a recipe for a cake that uses two eggs, and an altered gene as a mistyped recipe that asks for 12 eggs to be used. The recipe that uses 12 eggs will lead to a cake that will be different in taste or texture, or may not be edible at all.

Some altered genes have been linked to contributing to the disruption of normal motor neuron processes in MND. Around 10% of people who develop MND have inherited an altered gene.

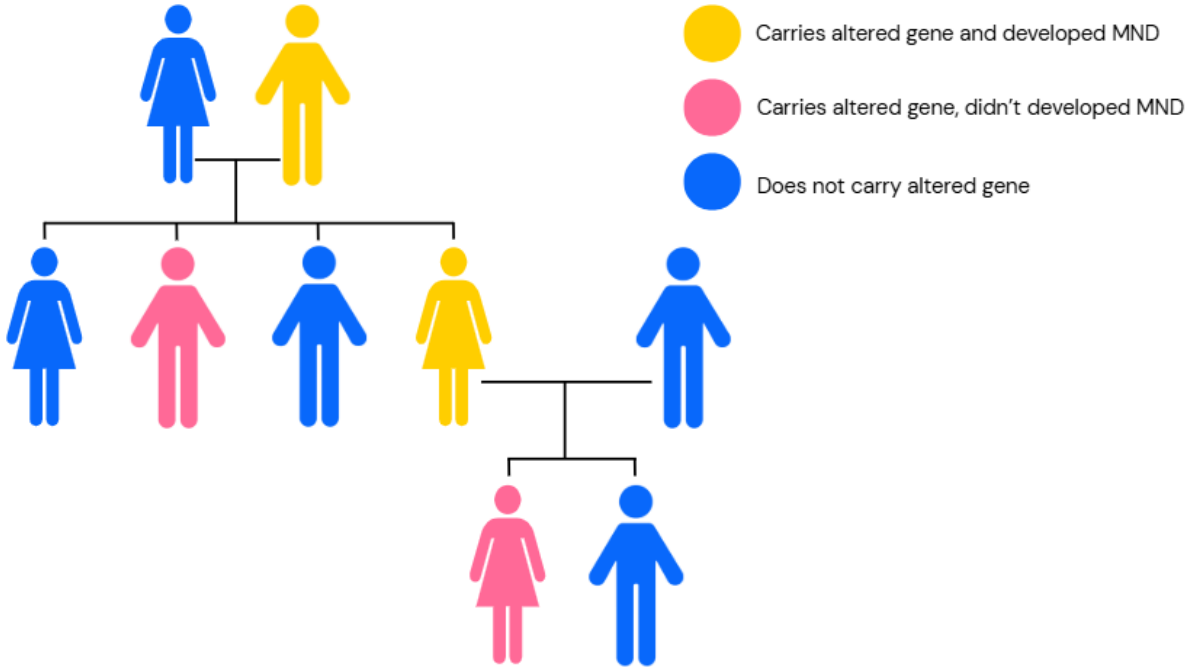
How do you inherit an altered gene?

Each person inherits two copies of each gene, one from their mother and one from their father. This includes genes that, when altered, can increase a person's chance of developing MND. If a parent carries one copy of an altered gene, there is a 50% chance they will pass this on to their biological children.

What effect does having an altered gene have?

Although we don't yet fully understand what causes MND, research suggests that there are several risk factors involved, such as genetics, a person's lifestyle, and environmental factors. **Importantly, a person inheriting a copy of an altered, known MND-related gene does NOT mean that they will definitely develop MND**, but it does increase their risk.

In genetics, this is known as reduced penetrance. A person may carry a copy of the altered gene but never develop MND. For example, a person who carries the altered gene may not develop MND, while a parent and grandparent, who also carried the altered gene, did. Example below:



What genes are linked to MND?

Alterations in over 40 genes have been identified, which are linked to MND. The four most common genes are:

- C9orf72
- SOD1
- FUS
- TARDBP

Can I get tested for MND-associated genes in Scotland?

If you are a person in Scotland living with MND, genetic counselling is available if you have a strong family history of inherited MND or if you display signs of cognitive impairment at diagnosis. You can ask your neurologist for a referral.

If you are a person in Scotland with a strong family history of inherited MND, you can be referred by your GP for genetic counselling.

In both cases, individuals may be referred to a regional genetic service, where tests will be carried out, although genetic testing after a referral is not guaranteed.

DNA banking is also sometimes made available if people do not want to know their genetic status now but would like the option of making that information available to others after they have died (for instance, children who might be worried about their own risk and would like to know about parental status).

Where can I find out more information?

If you have been diagnosed with MND and would like further information on genetic testing, you are best to speak to your Neurologist who will be able to advise you. If you have not been diagnosed with MND, but have concerns or a family history, you can speak to your GP.

A 2015 MND Scotland and Chief Scientist Office of Scotland funded project, carried out by Dr Danielle Leighton, mapped the genetic landscape of MND in Scotland. You can read the research paper from this study here - Genotypes and phenotypes of motor neuron disease: an update of the genetic landscape in Scotland - PubMed.

There are some research studies which focus on these inherited gene changes. You can find information on current open research studies and clinical trials you may be able to take part in here - UK MND Clinical Studies Group - Clinical research.