1: Motor neurone disease (MND)

This section provides basic facts about motor neurone disease (MND) and its diagnosis.

The following information is an extracted section from our full guide *Living with motor neurone disease*.

All of the extracted sections, and the full guide, can be found online at: [www.mndassociation.org/publications](http://www.mndassociation.org/publications)

The full guide can be ordered in hardcopy from our helpline, MND Connect:

Telephone: **0808 802 6262**
Email: **mndconnect@mndassociation.org**
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What is MND?
A French doctor called Jean-Martin Charcot first described motor neurone disease (MND) in 1874.

The term covers a group of related diseases that attack the motor neurones (sometimes referred to as motor neurons). These are the nerves in the brain and spinal cord that control how your muscles work. This means that messages gradually stop reaching muscles, leading to weakness, stiffness and wasting.

MND is a progressive disease, which means symptoms get worse over time. It can affect how you walk, talk, eat, drink and breathe. In some cases, it can also affect how you think and behave.

However, not all symptoms happen to everyone and it is unlikely they will all develop at the same time, or in any particular order.

“I hadn’t a clue what motor neurone disease was.”

Although MND is life shortening and there is currently no cure, symptoms can be managed to help maintain the best possible quality of life.

See Section 2: Symptoms and management.
Who does it affect?
Studies of MND indicate that:
• MND is a disorder which can affect adults of any age, but usually when they are more than 40 years old
• it is most common in people aged 55–79 years
• six people per day are diagnosed with MND in the UK
• it affects up to 5,000 adults in the UK at any one time.

What does it affect?
Your nervous system is made up of two main groups of nerves:
• nerves controlling the senses, such as touch and sight, known as sensory neurones (not generally affected by MND)
• nerves controlling the way our muscles move, known as the motor neurones (affected by MND).

The motor neurones are split into two groups:
Upper motor neurones: which run from the brain into the spinal cord carrying instructions to the lower motor neurones. Upper motor neurone damage tends to result in weak and stiff muscles.
Lower motor neurones: which run from the spinal cord and control muscles using the instructions received from the upper motor neurones. Lower motor neurone damage tends to result in weak and floppy muscles, and a twitching sensation that ripples under the skin (known as fasciculation).

With MND, the motor neurones become damaged and can no longer carry messages to the muscles. As this damage worsens, it can cause loss of mobility and movement, and difficulties with speech, swallowing and breathing.

The muscles first affected tend to be those in the hands, feet or mouth.
Up to half of people with MND experience changes to their thinking and behaviour.
In most cases, the changes are mild, but may be more severe for a small number of people.

See Changes to thinking and behaviour in Section 2: Symptoms and management.

MND does not usually affect the senses, bladder and bowel, or sexual function.
The effects of MND can vary enormously from person to person. Each individual will have a very different experience, from symptoms and disease progression, to the length of survival time after diagnosis.

How is MND diagnosed?
MND can be extremely difficult to diagnose for several reasons:
• it is a comparatively rare disease
• it can take time for someone to see their GP, as early symptoms, such as clumsiness, mild weakness or slightly slurred speech may have been thought due to other causes
• the disease affects each individual in different ways, as not all symptoms may be experienced or appear in the same order
• there is no test to prove someone has MND, although testing is used to eliminate other potential conditions.

"After nearly a year of tests, visits to my GP and hospital, MND was finally confirmed."

Tests
If your doctor thinks you may have a neurological problem, he will refer you to a neurologist at your local neurology department.

You may then need a series of tests. You normally attend these as an outpatient, but in some cases you may be required to spend a short stay in hospital.

Clinical examination: helps a neurological consultant recognise signs and determine which tests are appropriate, depending on your symptoms.
**Blood Tests:** look for a rise in a substance called creatine kinase. This is produced when muscle breaks down. It is sometimes found in the blood of people with MND, but may indicate other medical conditions.

**Electromyography (EMG):** is sometimes called the needle test, as fine needles record natural nerve impulses within certain muscles. When muscles start to lose their nerve supply, this can be detected by abnormal electrical activity, even if muscle activity is as yet unaffected.

**Nerve Conduction Tests:** apply an electrical impulse through a small pad on the skin to measure the speed at which nerves carry electrical signals.

**Transcranial Magnetic Stimulation (TMS):** measures the activity of the upper motor neurones to assist diagnosis.

**Magnetic Resonance Imaging (MRI) scans:** involve being placed in a cylinder-like machine to take internal images of the body. These help to rule out conditions such as stroke, Alzheimer's disease, Parkinson's disease, multiple sclerosis, tumours and trapped nerves, as well as injury to spine and brain.

**Other tests:** may be requested by your neurologist if their findings indicate they could be useful.

**Are there different types of MND?**

There are four main types of MND, each affecting people in different ways. However, these can have overlapping symptoms, which make it difficult to give an exact diagnosis.

The most common form is amyotrophic lateral sclerosis (ALS). Although other forms may be diagnosed, it is possible they will progress into the more typical form of ALS.

**The following definitions talk about life expectancy, so you may not want to read any more at this stage. If so, please move to the heading What causes MND?**

**Why include life expectancy here?**

Any information or discussion about life expectancy is likely to be upsetting. However, MND is a complex and challenging condition. This means lots of decisions may need to be made about future care, treatment of symptoms, and practical and financial matters. It can help to understand how the types of MND are likely to progress. This makes it easier to make informed and timely choices when planning ahead.

Life expectancy for each type of MND is based on average expectations from clinical studies. It is not an exact figure, as the speed at which the disease progresses can be rapid for some and slower for others.

**Amyotrophic lateral sclerosis (ALS):** is the most common form, involving both upper and lower motor neurones. It is characterised by weakness and wasting in the limbs, muscle stiffness and cramps. Someone may notice they are tripping when walking or dropping things. Average life expectancy is between two to five years from onset of symptoms.

**Progressive bulbar palsy (PBP):** affects only a small proportion of those diagnosed with MND, and involves the upper and lower motor neurones, particularly those linked to the bulbar regions in the early stages (muscles of the face, throat and tongue). Symptoms may include slurring of speech or difficulty swallowing. Average life expectancy is between six months to three years from onset of symptoms.

**Progressive muscular atrophy (PMA):** affects only a small proportion of those diagnosed with MND, mainly causing damage to the lower motor neurones, particularly those linked to the bulbar regions in the early stages (muscles of the face, throat and tongue). Symptoms may include slurring of speech or difficulty swallowing. Average life expectancy is between six months to three years from onset of symptoms.

**See Further information at the end of this section.**
Primary lateral sclerosis (PLS): affects only a small proportion of those diagnosed with MND, damaging only the upper motor neurones. This causes weakness in the lower limbs, although some people experience clumsiness in the hands or speech problems. Reflexes can become exaggerated. Average life expectancy may be more than 10 years. An email support group exists for this rare form. See Further information at the end of this section.

Kennedy’s disease
Kennedy’s disease is a rare neurological disease, causing increasing weakness, wasting of muscles and hormonal changes. It is not a type of MND, but can be confused with MND at diagnosis.

Most people with Kennedy’s disease develop symptoms at 30–60 years old, but it can appear when older or younger. There is no known cure, but most people with the disease live an average life span. Symptoms can be managed to improve quality of life.

Kennedy’s disease is caused by a genetic mutation, which can be diagnosed through gene testing. Usually, only men show symptoms, but women can carry the genetic mutation and in rare cases may develop symptoms. The MND Association offers support if you have Kennedy’s disease and this guide may be useful where symptoms are similar. See Further information at the end of this section for details about our information sheet on Kennedy’s disease.

What causes MND?
It is still not possible to give a clear answer about the causes of MND as each individual may be affected by a different combination of triggers. However, when you are being diagnosed, a neurological consultant will probably ask you about any family history of MND or frontotemporal dementia (FTD). If not, it is worth asking the consultant to discuss family history, as this may help determine if an inherited gene is one of the likely factors.

MND with no apparent family history:
Most cases of MND occur with no apparent family history of the disease and the precise causes in these instances are not yet known. Multiple genetic and environmental triggers are thought to be involved, with genes playing a small role. The environmental triggers may be different for each individual, so there is no simple way of identifying what may have played a role in the onset of the disease.

MND where there is a family history:
In a small number of cases, there is a family history and the genetic input is more significant. Where this occurs, the disease is caused by a mistake in the genetic code which can be passed down, although other triggers may still be necessary for the disease to emerge.

If you are concerned about the possibility of a family history of MND and what that could mean for those close to you (in terms of inheriting the genetic code), you may wish to seek genetic counselling. Although sensitive to the emotional aspects of the situation, genetic counselling is not a form of psychotherapy. A genetic counsellor explains the facts as clearly as possible, and gives you accurate information on the implications for your family.

This will include information about options such as genetic testing, to help you make up your own mind if this is a choice you wish to make. Some genetic testing is possible, but not everyone with a family history would benefit. Currently, testing is only available for four of the genes that play a part in inherited MND and results are not necessarily conclusive.
Choosing to be tested can be a very difficult decision, as it affects the wider family. We would advise genetic counselling from a neurological expert experienced in MND. In the first instance talk to your neurological consultant for advice.

For more information about inherited MND and genetic counselling see Further information at the end of this section and our research sheets on inherited motor neurone disease.

Research into MND

There has been an acceleration of world-wide research into the disease and its causes, including projects funded by the MND Association. As a result, our understanding of MND and the way motor neurones function is constantly advancing. You can keep up to date with latest findings through the research pages on our website, at: www.mndassociation.org/research

“ I am optimistic there will be a cure one day. It’s a challenge for the 21st century and I think science will find a way.”

Some research projects and clinical trials need the participation of people with MND, and occasionally their families. We know how keen some people are to help in this way and have developed a research list to help record this interest. Find out more at: www.mndassociation.org/researchlist or contact us to see if you meet the qualifying criteria: Telephone: 01604 611880 Email: research@mndassociation.org

Key points

- MND affects the motor neurones which we use to control movement. It does not usually affect the senses.
- There is no single test for MND. Testing is used to rule out other conditions.

Further information:

We have a range of numbered information sheets, including:

1A:   NICE guideline on motor neurone disease
2B:   Kennedy’s Disease
2C:   Primary lateral sclerosis (PLS)
2D:   Progressive muscular atrophy (PMA)
9A to 9C: our range of sheets on thinking and emotions in MND

Also research sheet B:
Part one – Introduction to inherited motor neurone disease
Part two – Genetic testing and insurance
Part three – The options available when starting a family

Most of our publications can be downloaded from our website: www.mndassociation.org or you can order them from MND Connect, our support and information helpline:
Telephone: 0808 802 6262
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MND Connect can also help you locate external services and providers, and introduce you to our services as available, including your local branch, group, Association visitor or regional care development adviser (RCDA).

See Section 14: How we can help you.

Our research website pages:
www.mndassociation.org/research

Online forum:
http://forum.mndassociation.org hosted by the MND Association for you to share information and experiences with other people affected by MND.

PMA/PLS email support group:
if you wish to join the email support group for primary muscular atrophy and primary lateral sclerosis, please email your details to care@mndassociation.org
The MND Association would like to thank the Tesco Charity Trust, and the Evan Cornish Foundation for their support which has made the production of Living with motor neurone disease possible.

The MND Association has been certified as a producer of reliable health and social care information. www.theinformationstandard.org

For references and acknowledgements please refer to the full guide, Living with motor neurone disease.

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