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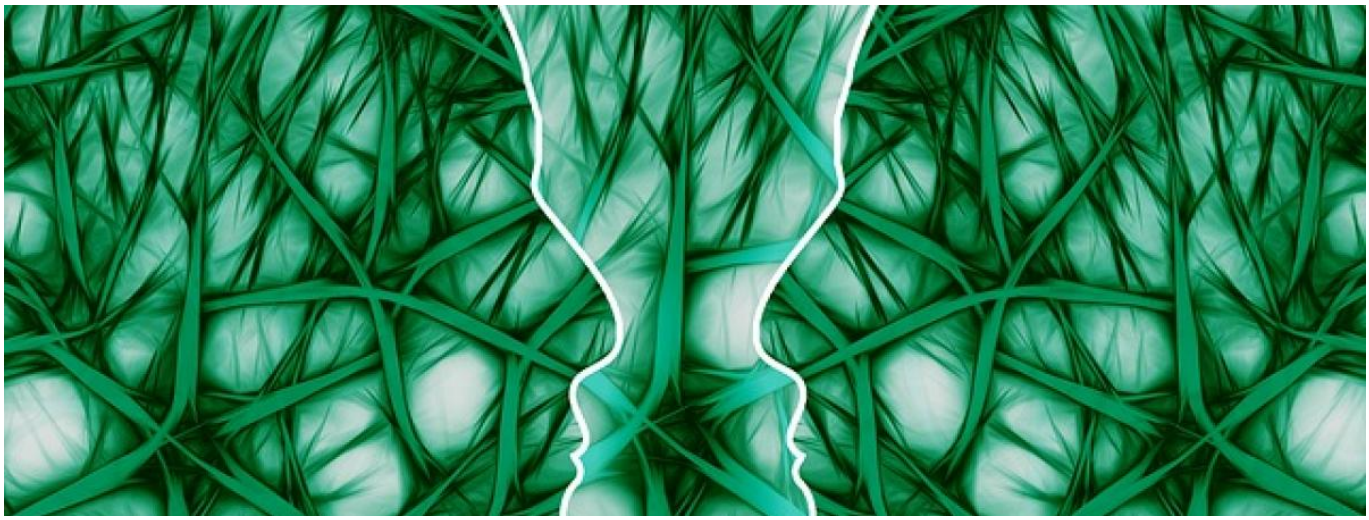

According to the Motor Neurone Disease Association, six people are diagnosed with the disease every day and six people will die each day. In the UK, there are an estimated 5,000 people living with motor neurone disease (MND). Helen Cowan speaks to Dr Andrew Douglas, consultant clinical geneticist at the Oxford Centre for Genomic Medicine, specialising in neurodegenerative disorders such as MND.

1. There have been no major breakthroughs in the 21st century and no new MND medication for 28 years.

True and false. It is true that there has only been one available drug (riluzole) for the treatment for MND in the past 28 years and at best it can only improve survival by a few months. However, at the same time there have been major breakthroughs in our understanding of MND, particularly in the discovery of many genes that cause familial cases of the disease. At the start of this century, there was really only one gene known (*SOD1*) where alterations in its DNA code had been found to cause MND. However, *SOD1* only accounts for a small proportion of affected people. Now there are well over a dozen definitive MND-causing genes and a couple of dozen more that are thought to cause MND with varying degrees of evidence. Knowing about these genes has started to allow the development of new targeted therapies.

2. ALS is the most common form of the disease.

True. "ALS" stands for amyotrophic lateral sclerosis, which describes the most common way that MND presents, with wasting away of the muscles and dying away of the motor nerve cells in the brain and spinal cord. In many

people this affects the limbs initially, while in others their speech and swallowing may be affected first and this is called "bulbar onset". Other types include "primary lateral sclerosis" (PLS) and "progressive muscular atrophy" (PMA). There are many other rarer ways and combinations of ways for the condition to start but the underlying process by which the brain and nerve cells die is essentially the same. However, terminology in the field of MND can be confusing. In the USA, the term ALS is used more as a general term to cover all forms of the disease, in the same way we in the UK use the term MND.

3. Every person with MND experiences the same symptoms.

False. There is a wide variety of symptoms and signs that can occur in MND. One person may first notice weakness in the legs, while another may initially notice arm weakness. Others still may have slurring of speech or very rarely MND may present with diaphragm weakness and difficulty breathing. The common feature of all these symptoms is that they do not improve but slowly get worse. Another important symptom that can be linked with MND is cognitive change that can affect language ability, behaviour and/or reasoning. Such changes occur in up to a third or more of people with MND and 15% of individuals may be diagnosed with frontotemporal dementia.

4. It can take up to a year for diagnosis.

True. Getting to the diagnosis does not always take as long as this but unfortunately the early signs of MND can be subtle. This can make the condition hard to diagnose early on and hard to distinguish from other neurological conditions. Furthermore, there is no single test that can confirm whether or not someone has MND and so the diagnosis relies on careful clinical assessment being undertaken by an experienced neurologist. Such assessment generally includes not only clinical examination but also imaging of the brain and spine by MRI to help exclude alternative causes of weakness. Blood tests and electrical testing of the nerves may also be used to help make the diagnosis.

5. No one knows what causes MND.

True. Despite the huge amount of knowledge we have gained about MND in recent years, we still do not really understand why the disease process starts at the age it does (usually in middle to later life) and why it only seems to affect the nerves that control movement and does not involve the sensory nerves that allow us to touch and feel, for example. Having said this, we do nevertheless know a number of factors that can increase the risk of MND. Apart from the genetic factors already mentioned, we know that men are a bit more likely to get MND than women, that smoking increases risk and also that exposure to certain chemicals and metals may play a role. Evidence now also seems to suggest that head injuries and regular intense anaerobic exercise may increase MND risk, although such risks are hard to quantify and must be weighed against the overall health benefits of regular exercise.

6. There are treatments to ease symptoms.

True. While no current treatment can stop MND from progressing, supportive and symptomatic treatments can help affected individuals have a better quality of life. Above all, a multidisciplinary approach to care is what is needed. Breathing difficulties are one of the major problems encountered in MND and careful assessment and monitoring of respiratory function can allow care teams to provide affected individuals with breathing aids such as non-invasive ventilation machine at an appropriate time. Weight loss and swallowing difficulties are also frequent problems and many people benefit from having a feeding tube (gastrostomy) placed in their stomach as and when this becomes necessary to maintain sufficient calorie intake. Adaptation of living arrangements, such as installing suitable equipment at home, and the use of modern communication aids such as voice banking for synthetic speech, can also help maintain a person's independence for longer.

7. Life expectancy is always short with MND.

False. Sadly, the most common clinical course in MND is a progressive deterioration of symptoms and the average life expectancy following diagnosis is only 2-4 years. However, some people with MND have a much slower progression rate of disease over many years. Perhaps the most well-known example of someone with a prolonged survival with MND was the physicist Stephen Hawking, who lived for a further 55 years following his diagnosis. It is generally impossible to know at the time of diagnosis how an individual with MND will progress. However, our increasing knowledge of the causes of this disease and the development of novel therapies based on that understanding gives some hope that in coming years we will have drugs that can at least slow down the disease. While a definitive cure remains hard to foresee, it may in time become possible to identify those at increased risk of MND and to offer preventative treatments to help stop the disease from starting in the first place.

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[1] https://www.readersdigest.co.uk/health/health-conditions/true-or-false-7-statements-on-motor-neurone-disease-mnd [2] https://helencowan.co.uk/../tags/disability [3] https://helencowan.co.uk/../tags/brain