

MND Matters: Episode 26: What is MND?

Intro

You're listening to MND matters, a podcast from the MND Association.

Helen

Welcome to MND Matters, brought to you by the MND Association and sponsored by Toyota GB and Toyota Financial Services. Hi, I'm Helen and I'm a regional fundraiser at the MND Association. We know that the awareness and publicity of MND is increasing. And this has been fueled by the tireless work of our members and the wider MND community alongside the huge increase in recognition of motor neurone disease in the media. From all of this incredible work, more and more people are now coming across MND for the first time, perhaps without any knowledge of what it is and what it means for those who are diagnosed. So in today's episode, I'm joined by regular host of MND Matters podcast, Nick, and we're going to be answering some basic and frequently asked questions about motor neurone disease.

Nick

Hi, thanks, Helen. I'm Dr. Nick Cole, the Head of Research at the MND Association.

Helen

Since then, it makes sense that we start with the question, what is MND?

Nick

Well, MND stands for motor neurone disease, and it's a fatal rapidly progressing disease that affects the brain and spinal cord. It attacks the nerves that control movement so that our muscles can no longer work. These nerves or motor neurons are like the electrical wires that send a signal along them to request muscles to move. And without that signal, the muscles can't be moved. And this causes the disease and motor neuron diseases and is an umbrella term for several diseases. One of which is ALS, which stands for amyotrophic lateral sclerosis, which is the medical term for it. So in the US and other countries they actually call MND, ALS, whereas in the UK, we call ALS MND and MND is an umbrella term for several of those motor neurone diseases.

Helen

What other types of motor neurone diseases are there?

Nick

Well, ALS is the most common form of MND and counts around about 90% of cases. And the other forms are a bulbar onset MND called progressive bulbar palsy, PBP, and progressive muscular atrophy PMA that affects only a small proportion of people relatively, and primary lateral sclerosis PLS, which is another rare form of MND, causing mainly weakness and stiffness that begins in the lower limbs, and

those four different types of MND. They all progress at different rates, and the symptoms first start in different areas, but ultimately, they're all motor neurone diseases.

Helen

What are the early signs or the first symptoms that somebody might notice of MND?

Nick

Well, the symptoms in people affected by MND are different. In every person, there's a range of different symptoms that can first be noticed, for example, some loss, muscle weakness, or loss of muscle mass, difficulties in movement and the ability, perhaps muscle cramps, stiff joints, people may notice speech and communication problems, so affecting how you speak, we estimate that around 80% of those living with MND will experience communication difficulties, including for some, a complete loss of voice, swallowing difficulties, difficulty with weak coughing and breathing problems. And in some people, there are some changes in emotion. So a varied combination of symptoms. And as I say, in each person, MND tends to start differently.

Helen

How might symptoms then progress from those early signs?

Nick

So usually, those symptoms will progressively get worse over time. And it can be slow in some people, and fast in others. So one of the curious things about motor neurone disease, that is the rate of progression, as it's called, the symptoms can happen faster in some people and slower in others.

Helen

How can we diagnose MND?

Nick

MND, currently is very difficult to diagnose. It's almost more a process of elimination. So discounting other diseases before somebody gets that diagnosis, what we really need is a biomarker, an indicator, like a blood test. So for example, in diabetes, you can test the level of insulin. And a blood test to help with diagnosis of MND is really important not only to speed up that diagnosis, because once we get effective therapies they need to be given to people as early as possible in the disease, but also they help in readouts for clinical trials. So there is considerable work looking for biomarkers in motor neurone disease and we think we have one now called neurofilament and some really comprehensive work has gone on to look at that. And it's part of the structure of the motor neurone. So as they break down, the neuro filament is released into the bloodstream and spinal fluid and that can actually be measured to give a readout of the level or progression of motor neurone disease and of course and to answer your question, in terms of diagnosis, a high level of neurofilament could be an indicator that somebody has motor neurone disease.

Helen

What isn't affected when somebody is diagnosed with MND?

Nick

Usually the senses are not affected in people with MND. In a small proportion of people, there are some changes in the way that people think. But generally, people will still feel all the things they normally feel physically and be the same person inside. Whilst all of these physical changes are happening to their bodies.

Helen

I know the MND Association funds, an awful lot of research do we know what causes MND?

Nick

This is one of the really difficult things about motor neurone disease and neurological diseases in general, it's really difficult to see inside a person to see their motor neurones and their nervous system. So in terms of what actually causes MND, we don't exactly know what the causes are. We know that it's a combination of genetic, environmental and lifestyle factors. One analogy is to think of grains of sand on a scale. And if you're exposed to a certain number of different factors, it can tip the scales towards showing MND symptoms.

Helen

Do we know is that indeed hereditary? Is it genetic? Is that a reason why some people get MND?

Nick

In some cases around about five to 10% of all motor neurone disease, there is Motor Neurone Disease running in families. And this indicates that there's a genetic cause. And indeed, thanks to research, we've been able to identify genes that can cause inherited MND. So, as I say, around about five to 10% of all motor neurone disease, there is a genetic basis. And if we go back to that scales analogy, you can think of those gene changes or those particular genes, tipping the scales towards showing motor neuron disease symptoms. It's really important to emphasise though that just because somebody may have a genetic change, they won't necessarily show the symptoms or develop motor neurone disease you need, we think those other combinations of lifestyle environmental factors to tip you into that state where you're showing symptoms of motor neurone disease and develop the disease.

Helen

If somebody is diagnosed with MND, what is the average life expectancy?

Nick

The life expectancy of people with motor neurone disease can vary greatly. Some people live for a considerably long time with the disease. But in the majority of cases, around about half of the people diagnosed with MND will, unfortunately die within the first few years after diagnosis. We know that the lifetime risk of developing MND is around about one in 300. So one in 300 people will develop motor neurone disease in their lifetime. In the UK, six people are diagnosed with MND every day. And unfortunately six people die with MND every day.

Helen

Wow, those are really devastating facts about MND. And it's exactly the reason why the MND association is fighting back for all those living with and affected by this cruel disease. So now we know kind of how it affects people living with MND. But how does it affect those around them, the people that love them, the families and carers.

Nick

MND is quite a devastating diagnosis, it's very difficult to to put myself in that position. From what I've seen in my experience of working with people living with and affected by MND is that is the constant changing. So as the symptoms progress the needs and and care for somebody with motor neurone disease is constantly shifting. So it's not something that you can ever really get used to and start to, to live within in a sort of steady state. There's lots of changes that happen all the time. People are incredibly generous in giving their time when they're going through motor neurone disease to research projects and none of this research and these discoveries can happen without that selfless commitment of people doing that while they're going through the disease. It's not for me really to speak about how what it's like for for those families as I've never experienced it personally but um, it's it is amazing the strength people have and that's really reflected in how our community as well is so united in a fight against MND and the lengths people go to to, to raise money and to look after each other

Helen

Yeah, I think if you if you've loved somebody if you if you've known somebody with MND, you become be, you know, fiercely sort of dedicated to fundraising or supporting others because you've experienced it and it's through that fundraising and through kind of supporting each other that we'll find that back you're potentially in and it's the a world free from them, MND.

Nick

I think so. And it's important that people have a way to fight back against the disease. And I think that's, that's one of the real drivers for that.

Helen

Okay, so a lot of people will will know MND, from Stephen Hawking, who famously had MND was diagnosed in his 20s and lived for over 50 years with MND. So how, how was he different?

Nick

Professor Hawking had MND for a long time. It's really been put down to timely coordinated multidisciplinary care, and the use of key interventions, such as tracheostomy. But there might also be genetic and biological factors that are responsible for that slow progression. And that's one of the things that research is really interested in. Are there protective genes and factors, for example? And can we identify what they are? And can that be used to slow down the progression of other people with motor neurone disease? So really, the more we understand about the disease and the biology of it all, the more we can target particular mechanisms to to have effective therapies.

Helen

One thing I'm asked quite regularly is is MND, more regular in men than women? I suppose there's lots of celebrities that have been diagnosed, that are men. So what's the prevalence of MND in different communities?

Nick

There is a slight difference in the numbers of men with motor neurone disease versus women, slightly more men get motor neurone disease than women. We're not exactly sure why that is at this point. And that's hopefully something will come to understand further online. MND doesn't discriminate. It can affect anyone, from any community. Any background, we do know that the older a person is, the more likely they are to get MND, and MND is rare in teenagers and young people. But there are cases certainly of young people and teenagers actually getting MND.

Helen

More research is needed, isn't it to kind of answer those, those questions of the trends that we see.

Nick

That's right. There's lots of unknowns about motor neurone disease. And we're we're working very hard to understand more about the biology and the causes, and how we can develop effective therapies to slow it down. And hopefully, stop MND.

Helen

Talking of therapies than it is is there currently a treatment for MND?

Nick

Unfortunately, in the UK, at the moment, there's only one treatment that's licensed for motor neuron disease called riluzole. And this only has a modest effect on people's life expectancy. So really, we don't have any effective treatments at the moment. The good news is that there are quite a few clinical trials and lots of positive research projects going on. And we're quite hopeful that some of these drugs and compounds that are being tested at the moment are going to lead to effective therapies in the future.

Helen

Here's a hard question for you, Nick. I'm putting you on the spot a little bit. How far do you think we are from an effective treatment then for cure for MND?

Nick

That is a difficult question. But I'm always happy to answer that. I mean, it could be tomorrow, it could be a long time in the future. But the good news, again is that there's more research going on right now than there ever has been before. And we're really on the cusp, I feel of some major breakthroughs in MND. There's one particular genetic therapy, called Tofersen which is showing considerable promise, we think and that that's effective in a genetic form of motor neurone disease caused by a gene called SOD one, which was the first gene to be discovered, associated with MND. And this has shown some real promise. It's important to note this, this will only be effective in around about 2% of people with MND. But it's really the first cab off the rank as it were, which has shown an effect. And there are lots of

other therapies, gene therapies using the similar principle that are now in the pipeline and being worked on and a great interest by pharma in, in putting money into MND to understand how these therapies could work. So some of these therapies are actually going to be we hope, if they work, there'll be effective in a larger proportion of people with MND, so not those with just the specific gene mutations. So lots of good things, really, in the pipeline. So in terms of when I can't say when, but I think it certainly feels like it's going to be sooner than it's ever been before. And, yeah, I wouldn't like to put a time on it. But if I was absolutely forced, I'd say was certainly within the next five years or so we should have at least one effective therapy from MND. My fingers are very much crossed with that.

Helen

Wow, wow, that that kind of makes me a little bit speechless. So as a podcast host that that's a difficult position to be put on there. But I think that just highlights the importance of fundraising and all of our supporters that have, you know, gone out there done something silly or put themselves through a marathon or bake some cakes, because it kind of does prove that if we can fund the research, we can get those answers and we can get closer to that effective treatment. That's unbelievably exciting.

Nick

That's absolutely right. I mean, none of this research and this understanding that we're developing can be done without funding. I mean, it's been said a few times that MND isn't incurable. It's just underfunded. And every single penny that goes in to help towards research is finding those discoveries and is absolutely and it's shown in the results that have come out recently, that it's really moving us towards and closer and closer to an effective treatment.

Helen

Okay, well, I think I'm, I'm gonna leave it there with the questions and leave it on that hope and I really look forward to all of the research advances over the next five years. Thank you so much, Nick, for answering every question that's been thrown at you. It I'm sure. It's kind of given lots of people an insight into now what MND is, it's not just three letters, it's motor neurone disease. But if anyone's listening if you have any further questions about MND or would like some more information about how you can get involved to support the 5000 people living with MND today in the UK, please do go onto our website. MNDassociation.org. And if you need support, please call our MND Connect helpline on 0808 8026262.

Outro

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