MND – Our Endless Nightmare

Dr Justin Yerbury

Have you ever had a dream where no matter how hard you try it is impossible to move your body, and when you call out for help you don’t make a sound? This is a frightening experience that people wake from, in a pulse racing panic. Imagine now that this has happened to you, but you could not wake up from the dream. This is how a patient with Motor Neurone disease (MND) lives through the last stages of their life.

MND is characterised by a rapid loss of the neurones that control voluntary muscles. Sufferers progressively lose the ability to use various muscles in the body, leading to an inability to walk or pick up objects; even feeding oneself becomes impossible. Ultimately the act of breathing becomes too onerous for the already frail body, and sadly the disease invariably ends in death within 1-5 years of diagnosis. These are confronting thoughts, but these are the realities faced by around 1400 Australians with MND and thousands more, such as carers, families and friends, constantly living with its impact. On average, at least one person dies from, and another is diagnosed with MND, each day in Australia.

In the Illawarra one in every 12,000 people is affected by MND. The cause of most cases of MND is unknown and only 10% of all cases are inherited with certainty. Even though several specific genes have now been linked to the inherited disease, a large proportion of inherited or ‘familial’ cases are still not linked to a particular gene. Epidemiological studies have not identified clear causative agents for the remaining 90% of sporadic MND. However, we do know that MND affects males and females at the same rate and prevalence is not based on ethnicity or demographic variables apart from age. Typically the average age of onset is about 50 years old, however, cases have been reported in adults as young as 21. Environmental factors are likely to play a role in disease onset as researchers have found increased incidences among various populations such as the Chamorro people of Guam (where in the 1950s MND rates were 50 times higher than expected), in Gulf War Veterans and professional football (soccer) players in Italy.

Currently, there is no effective therapeutic to stop progression or prevent onset of MND. Rilutek, a drug approved for use in MND treatment, offers for most patients only a very modest effect generally described as a three-month life extension. MND is sometimes referred to as an orphan disease because it has not been “adopted” by the pharmaceutical industry. Sadly, the relatively small number of patients worldwide means it provides smaller financial incentives to produce and market new drugs.

At the Illawarra Health and Medical Research Institute work is in progress to determine the underlying mechanisms that cause MND, knowledge that will help fight the disease. One current project is examining the role of damaged proteins in motor neurone death. Like most biological molecules proteins are constantly being made, used, and disposed of in neurones. Tight regulation of this process means that if a protein is damaged in any way it is normally disposed of rapidly. A characteristic trait of motor neurons in MND is that proteins accumulate into deposits, which are essentially large ‘junk piles’ that cells are unable to dispose of. We hope to discover how these ‘junk piles’ are killing cells and believe this work may identify processes that underpin MND.

These studies, currently underway in our Illawarra region, may take us one step closer to finding a cure for this devastating and currently incurable disease.

Justin Yerbury is a research fellow at the University of Wollongong and a member of the Healthy Ageing Research Theme of the Illawarra Health and Medical Research Institute (IHMRI).