

Motor neurone disease (MND): a personal perspective

Roderick A Mackenzie

“ROD, ARE YOU SURE IT’S MND?” It was James Lance, my former Professor of Neurology (now Professor Emeritus, University of New South Wales), on the telephone. He had just received the letter I sent to all my close friends and colleagues when the diagnosis was confirmed. Jim had always emphasised the importance of leaving no stone unturned before accepting a diagnosis of incurable disease, both in his book on headache — which inspired me to do neurology — and during my subsequent clinical training. However, I had to tell him there was no doubt. Four months previously, I had experienced the sudden dramatic onset of widespread muscle fasciculation, without definite weakness, and, after four weeks of rising anxiety, I took my concerns to a colleague. Although at that stage he reassured me that he had found no definite abnormality, I was concerned that during the examination I had not been able to support my weight on my left leg.

I then entered a period of uncertainty, during which I could not discuss my fears with family or friends. Three weeks later my colleague found measurable muscle atrophy and hyperreflexia, and electromyography documented fasciculation and early muscle denervation. Magnetic resonance imaging and a second opinion finally confirmed the diagnosis of MND.

When I went to see David Burke, my then Professor of Neurology (Institute of Neurological Sciences, Prince of Wales Medical Research Unit, Sydney, NSW), to tell him my news, he tried to focus his mind on practicalities. As a young registrar he had supervised my research for an MD thesis and now, as my Chief of Neurology, he would have to arrange my replacement as Visiting Medical Officer and Director of the Comprehensive Epilepsy Service at Prince of Wales Hospital. However, for some moments all he could say was, “Rod, we go back a long way...”

It is now 16 months since the onset and there has been significant progression of weakness and spasticity. I can look back on my experience of MND thus far and make some observations.

Diagnosis

The final confirmation of the diagnosis came almost as a relief after the period of uncertainty and false hopes — certainly the worst four months of my life. Once the diagnosis was confirmed, it was as if a great weight had been lifted from my shoulders and I was able to face the challenges ahead. The hardest task was breaking the news to

my wife and each of my four children, but then they were able to provide much-needed emotional support. I was also able to begin the complicated process of selling my practice and disengaging myself from all my medical commitments. A lot of friends and colleagues were surprised by the speed and completeness with which this was achieved and still ask me if I miss medicine. On looking back, I can honestly say I have no regrets about this course of action. Freeing myself from the day-to-day concerns of clinical practice has allowed me to make up for lost time, especially with my family, and to “smell the roses”. It has also made easier the transition from doctor to patient.

This experience has reinforced my long-held view that one must be completely frank with patients about the diagnosis and prognosis of terminal illness. The neurologist who has assessed the patient and confirmed the diagnosis of MND is in the best position to break the news, and then to discuss the myriad issues that will come up. These include a frank discussion of prognosis, the manner of progression, symptoms to be experienced, the role of exercise and drugs and the possible need for assisted breathing and feeding in the future.

Aetiology

Although most cases of MND are sporadic, up to 10% are familial and may be associated with a mutation on the superoxide dismutase gene.¹ I was relieved to find that this test was negative in my case, reducing the chances of other family members being affected.

MND patients have a higher than normal incidence of previous paralytic poliovirus infection. In 1951, when aged 5 years, I suffered a febrile illness which was associated with diplopia, recurrent seizures and coma and followed by three weeks of limb weakness. Although I made a complete recovery without ventilatory support being needed, this was thought to be poliovirus encephalomyelitis and I suspect that this illness may have played a role in my MND.

Ongoing care

The general practitioner should, as always, coordinate care, but the possibility of this diagnosis should always lead to a referral to a neurologist. One or both of these practitioners might feel that one of the specialist MND clinics, now available at several of the teaching hospitals, is the best place for making the definitive diagnosis and providing the ongoing care. These clinics provide an expert diagnostic service, and they can monitor progress and arrange referral to a number of ancillary services. When necessary, they arrange provision of aids and can refer patients for advice on respiratory support and gastrostomy feeding at the appropriate time.

My GP put me in touch with the MND Society of New South Wales, a registered charitable, not-for-profit organisation which provides information, outreach support and

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access to disability aids and equipment. I have had a home visit from my designated outreach worker to discuss the diagnosis and prognosis, and my current and future needs and those of my wife as future carer. I have since become a board member of the society to assist them in their admirable work.

Role of exercise

As a neurologist I had always advised my patients to exercise only to the onset of muscle pain or weakness and then to cease, as exercise beyond this point was thought to be unhelpful and possibly harmful. However, as a patient, I found that, if I continued to exercise despite these symptoms, over the next few days the muscle pain and stiffness resolved and the weakness improved, even though it did not recover to previous levels. I was able to maintain a program of exercise with weights, stair-climbing and walking for two to three hours a day for 12 months before increasing weakness and spasticity forced a gradual reduction and finally cessation of these activities. This program was so successful that it is spasticity rather than weakness that is now forcing me off my feet.

Role of drugs

The American Academy of Neurology (AAN) Practice Parameters for ALS² represent a significant advance in formulating evidence-based guidelines for the care of people with MND and should be consulted when treatment options are being considered.

Riluzole is the only drug which has been shown to prolong survival in MND. Two large randomised trials³ suggest that it improves survival of MND patients by at least three months. I began taking the drug as soon as the diagnosis was confirmed and have had no side effects. It costs \$700 per month on private script, but it is currently being considered for listing on the Pharmaceutical Benefits Scheme.

Baclofen is essentially the only muscle relaxant available, as the alternative, dantrolene sodium, is associated with unacceptable side effects (asthenia and muscle weakness). I have been taking baclofen with only modest benefit, but no side effects.

Oxandrolone is a synthetic anabolic steroid, and a pilot trial of this drug over 12 months involving 12 MND patients found that the most severely affected muscles underwent little or no further deterioration, while less affected muscles continued to deteriorate.⁴ I began taking this drug (cost, \$1400 per month) three months ago, but unfortunately this has not prevented progression of weakness of my left leg, nor of other, less affected muscles. I have therefore ceased taking the drug.

Minocycline, a semi-synthetic tetracycline derivative, has been found to improve survival in a mouse model of MND,⁵ and I have been taking it with no side effects.

Progression

In MND, respiratory muscle strength is a strong predictor of survival, and death usually results from respiratory failure. Non-invasive ventilation improves quality of life and possibly survival.⁶ The AAN recommends commencing non-

invasive ventilation when vital capacity is less than 50% predicted. Recent work⁷ suggests that measurement of sniff nasal inspiratory pressure is more predictive of benefit, and that non-invasive ventilation should begin when sniff nasal inspiratory pressure is less than 60% predicted. I am monitoring both vital capacity and sniff nasal inspiratory pressure and will make a decision about non-invasive ventilation, and the option of gastrostomy feeding, closer to the time these may be necessary. Meanwhile, I am content to take one day at a time. As someone said, "Every day you're breathing is a good day".

Life issues

I now realise that the most important entities of my life are my family and friendships sustained over many years. Although my medical career defined my day-to-day existence and most others' perception of me, its significance pales in comparison. However, my knowledge of medical issues, such as those with which I am now personally faced, will hopefully help me cope with what lies ahead.

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