MND Podcast number one:

Presentation, diagnosis and clinical decision-making

Hello my name is Robyn Reid. I am the MND Shared Care Worker in the Southern Region of Melbourne. Welcome. This is a series of podcasts on Motor Neurone disease. This podcast may help you to understand Motor Neurone Disease and how its presentation and progression in the client will inform treatment decisions.

Every day we get out of bed and have breakfast, get dressed, drive or walk and go about our day. We do not have to think about the hundreds of voluntary movements involved in these activities. The muscles involved in speaking, breathing, moving, coughing, and swallowing occur without effort or without thinking about it. Imagine then the person with MND who notices the first signs of something not working in their body. They may stumble slightly on a regular basis. They may have some difficulty swallowing, they may fail to open that jar or container. They may not be able to button their shirt or may be short of breath. They may rationalise these symptoms away by saying “oh well I am getting older”.

MND is typically a disease that strikes men more than women and is more common in the 50-70 years old age group. However this disease can be found in people in their 20’s although this is fairly rare. MND has no cure. There is no remission and once function is lost it does not return. MND is a disease of losses: loss of mobility, loss of voice and speech, loss of identity and role, loss of employment, loss of a future and all the dreams associated with that. The disease moves at different rates affecting different parts of the body and in different patterns so there is no recipe for managing MND. No one person

with MND will be the same as another as each person is unique. However, in the end the cause of death for people with MND is respiratory failure.

The average life span is 3-5 years. When symptoms start the person may find it difficult to pin down a diagnosis. There is no definitive test. Diagnosis relies on things like medical history, an examination by a neurologist, CAT scan, Myelogram, MRI, Electromyography and blood tests. The person may not get the diagnosis for some time and this may mean that half their remaining life span is over before they know they have a life limiting illness. Some may experience rapid progression of their disease while others will have a slower decline. As symptoms progress it may be more difficult to make decisions so early discussions about the impact of the disease are important. Scientists are currently working on a blood test with biomarkers which could assist with diagnosis. Other neurological diseases need to be ruled out such as Multiple sclerosis and Parkinson's disease.

The person with MND will have symptoms that get worse over time. The different ways that the symptoms of MND occur and develop are called Phenotypes. Symptoms may start at one end of the body in the head and move down, or at the feet and move upwards. Symptoms may start in the middle of the body and move up or down, or start in the middle and move in both directions at once. The important thing about these patterns is that whatever part of the body is affected or about to be affected, will determine the treatment issues. Health professionals need to be able to predict and plan ahead for future needs such as wheelchairs or communication devices. The

client needs to be able to think about their life and the needs of their loved ones now and in the future.

Motor Neurone disease is a progressive neurological disorder in which motor neurones degenerate and die and voluntary muscles gradually weaken and waste. It is known as Amyotrophic lateral Sclerosis (ALS) and is sometimes called Lou Gehrig's disease. Sixty –five percent of people have upper and lower motor neurone involvement, twenty –five percent have Progressive Bulbar Palsy which starts in the head and affects speech and swallowing and moves down the body. Nine percent have Progressive Muscular Atrophy which can affect the arms and legs and cause a lack of ability to use limbs, called flail arm or flail leg. One percent of people have Primary lateral sclerosis which is a rare form with a much slower progression of the disease with life expectancy between 10-20 years. They may live for a decade or more before the disease is fatal.

Being told you have this disease is devastating because there is no cure. At the moment there is drug therapy with Riluzole which may extend the life of the affected person by a few months at best. People may respond with a range of reactions such as shock, sadness, anger, or relief at finding an explanation for their symptoms. They may feel hopelessness at the prospect of living with something that has no cure and will take away their independence. Sometimes there is denial about how serious the disease is and that it will end their life. There is often tension between health professionals attempting to anticipate the future needs of ongoing deterioration which cause further lack of function,

and the psychological and emotional readiness of the client with MND to come to terms with what is happening. There is always continued degeneration of neurones and further loss making it difficult for the client or their carers to find any emotional stability.

The cause of MND is unknown. 5-10 % of people have Familial AlS and will be able to identify someone in their family who has the disease. In 20% of this 5-10% group, there is a genetic mechanism known as mutations on the chromosome 21 superoxide dismutase gene, commonly called SOD1. However even if people can identify this gene for themselves, it is cannot predict the development of MND so genetic counselling has limited value. The vast majority of people have sporadic MND in which the disease develops for no apparent reason. The cause of MND may be a combination of aging, genetics, lifestyle and environmental factors but research has not identified specific or significant risk factors.

People with MND have their sensory system intact so will still experience pain and pressure. They are generally not incontinent although mobility issues may mean that without assistance with movement and clothing they will not make it to the toilet in time. They may also have bowel issues related to medication, nutrition and hydration. Fifty percent of people may experience changes in understanding,decision-making and paying attention. They may have trouble finding the right word, missing words or repeating words, on top of the mechanical difficulties of speech. They may have reduced ability to read emotions in others or to express their own emotions. There may also be

apathy, rigid thinking, impulsivity, and agitation as well as socially inappropriate behaviour. In most cases the cognitive impact is mild but as many as 5-15% may develop Fronto-temporal dementia. This has an impact on appropriate treatments as well as carer burden. Some of these changes may even occur before the physical ones. People with Progressive Bulbar Palsy may suffer Pseudobulbar affect in which they may not be able to control laughing or crying easily once it starts. Emotions may be exaggerated or different to those being felt. However, this is not necessarily indicative of cognitive change.

So in summary there are a number of types of presentation of Motor Neurone Disease with the most people having upper and lower motor neurone involvement where motor neurones die and voluntary muscles are affected. These are Amyotrophic Lateral Sclerosis, Progressive Bulbar Palsy, Progressive Muscular Dystrophy, Primary Lateral Sclerosis and Flail conditions. Where the disease is causing symptoms then these will determine the priority and urgency of clinical decisions. For example Bulbar palsy means that the person will experience swallowing and speech issues earlier than someone who is losing mobility. The person with Progressive bulbar palsy will need urgent assistance with future communication, swallowing and nutrition. The person losing mobility may need changes to their home for ramps and wheelchair access, and may take some time to experience speech and swallowing difficulties. In either case the disease is progressive and both will have respiratory issues at some point. Once swallowing is affected it becomes necessary to consider a Peg tube, which stands for (Percutaneous Endoscopic

Gastrostomy- which is a tube inserted into the stomach). This ensures adequate nutrition and hydration even though people can still take in food and fluids by mouth. People need enough respiratory function to have the surgery safely so this decision must be made sooner rather than later if chosen. Likewise if respiratory function gets worse then it may be necessary to consider Non- invasive Ventilation, which is a machine and mask to assist breathing. This improves quality of life and function. Not everyone will choose to have a Peg and NIV both of which will extend their life to some extent. These are some of the decisions the person must make as their disease progresses.

The person with MND is likely to have fears and questions about death and about how they will be cared for in the very final phase of the disease. The person with MND and their family may also have questions about when to go to hospital or a hospice, or whether they can stay at home for as long as possible, even until they die. It is important for these fears and questions to be discussed. Palliative care professionals can help with this. The community palliative care team should be involved and can provide support to both the person with MND and the family. Early referral is helpful.

One of the most common fears is that of the person choking to death. Although a person with MND may experience some choking episodes, it is very rare for this to be the cause of death. Some people with MND do die suddenly due to a sudden weakening of the respiratory muscles or chest infection, but

most people gradually deteriorate due to increasing respiratory weakness an eventual respiratory failure-even if they are using NIV.

I look forward to discussing more detail about various aspects of MND management in future sessions which will be available on the SMPPC website. I would like to take this opportunity to give my thanks to staff at Bethlehem Calvary health care and MNDVictoria for the support and training received in my role as MND SCW in the Southern Region. Please feel free to contact me should you wish to have an education session on MND for your agency or to receive a secondary consultation. You can contact me on: 0428264446 or email me at: Robyn.reid@smrpcc.org.au

Thank you.

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