

This Booklet has been reviewed and approved by the Australian Association of Neurologists. It was compiled from material written by Neurologists in the United States, United Kingdom and Australia (see acknowledgments at the end of the booklet), and is designed to be available on request as a supplement to the Leaflet "Motor Neurone Disease - Some Facts."

The Motor Neurone Disease Association of Australia is committed to working with the Australian Association of Neurologists, individual Neurologists and other professionals to improve the quality of services and outcomes for people living with Motor Neurone Disease.

What is Motor Neurone Disease?

Motor Neurone Disease (MND) is the name given to a group of diseases in which the nerve cells (neurons) controlling the muscles that enable us to move, speak, breathe and swallow undergo degeneration and die. Motor function is controlled by upper motor neurons in the brain that descend to the spinal cord; these neurons activate anterior horn cells (lower motor neurons). The lower motor neurons exit the spinal cord and directly activate muscles. With no nerves to activate them, muscles will gradually weaken and waste.

The patterns of weakness vary from person to person.

What are the symptoms?

Early symptoms are mild, and include problems with walking, holding objects due to weakness of the hand muscles, slurring of speech or swallowing difficulties due to weakness of the tongue and throat muscles. Emotional responses may be more easily triggered and the person with MND may be aware of laughing and crying more readily than previously. Cramps and muscle twitching are also a common symptom.

There is considerable variability between patients in respect of initial symptoms, rate and pattern of progression, and survival time after diagnosis.

How is it diagnosed?

The diagnosis of MND is often clinically difficult, and sometimes it is necessary to review patients for some time before the diagnosis becomes relatively certain. The family doctor may suspect a neurological problem, and organise referral to a

Neurologist. Several other neurological conditions resemble MND, especially in the early stages, and need careful exclusion.

The diagnosis can be assisted through a range of tests, including some which eliminate other conditions. Nerve conduction studies (NCS) and Electromyography (EMG) are often performed and may help in establishing the diagnosis. These studies involve analysing neural function by electrical stimulation of nerves (NCS) and recording muscle activity. EMG utilises a needle electrode which is inserted into various superficial muscles enabling analysis of their function.

What causes MND?

The causes of MND are not yet known, but ongoing research throughout the world is looking for causes.

There are many theories which include exposure to environmental toxins and chemicals, infection by viral agents, immune mediated damage, premature aging of motor neurons and loss of growth factors required to maintain motor neuron survival.

Is it hereditary?

Most cases of MND occur spontaneously. They are said to be sporadic - meaning occurring in scattered or isolated instances without clearly identifiable causes.

Some cases of MND are hereditary, with about 10% of cases having been shown to have a genetic link. In March 1993 mutations were identified on the superoxide dismutase (SOD1) gene on chromosome 21. These mutations are believed to be present in about 10 to 15% of the hereditary form of MND. Current research aims to understand how these mutations cause damage to motor neurones.

Motor Neurone Disease Associations in each State keep a close watch on research progress and findings and always have the latest information available.

Can other people catch it?

There is no evidence that MND is transmissible from person to person.

How common is it?

It is an uncommon, but by no means rare disorder. It occurs at similar rates in most countries of the world. It is estimated that there are presently about 1,400 patients in Australia.

What forms does MND take?

MND takes several forms. In some cases the symptoms are widespread from the onset, but usually MND begins in a localised fashion, affecting a single limb or aspect of motor function, becoming more generalised as the disease progresses. There is usually evidence of wasting and weakness in some muscles and twitching (fasciculation) of parts of the muscle may be visible under the skin. Fasciculation is

a sign of damage to lower motor neurons. Involvement of upper motor neurons results in stiffness of muscles (spasticity) and slowed movements, sometimes the muscles will spasm or spontaneously jerk.

Patients may have only lower motor neuron or only upper motor neuron signs early in the illness but typically develop a combination of the two as the disease progresses. Depending on the pattern of motor neuron involvement and the part of the body where the symptoms begin, the disease can be classified into four main types.

Amyotrophic lateral sclerosis (ALS) is the most common form, characterised by muscle weakness and stiffness, over-active reflexes and rapidly changing emotions. Upper and lower motor neurones are both affected; the limbs cease to work properly. ALS is the term most commonly applied to MND in the USA, where it is also known as Lou Gehrig's disease, after a famous baseball player.

Progressive muscular atrophy (PMA) is characterised by muscle wasting and weakness, loss of weight and muscle-twitching, and is caused by damage to the spinal (lower) motor neurones.

Progressive bulbar palsy (PBP), mixed bulbar palsy and pseudo-bulbar palsy are forms that involve the muscles of speech and swallowing. The nerves that control these functions are located in the bulb (the lower part of the brain), hence the term bulbar palsy (paralysis).

Primary lateral sclerosis, in which upper motor neurone damage results in stiffness and spastic paralysis of the limbs. This is a very rare form of the disease.

Other Diseases Affecting Anterior Horn Cells

Kennedy's disease is a disorder of motor neurones which is not Motor Neurone Disease. It is much more benign and is an inherited disorder affecting adult males. It causes slowly progressive weakness and wasting of muscles.

There are two very uncommon hereditary forms that affect infants and children.

Infantile spinal muscular atrophy (Werdnig-Hoffman disease), in which symptoms may be present at birth. Survival beyond the age of two is uncommon.

Juvenile spinal muscular atrophy (Kugelberg-Welander disease), in which onset is usually between the ages of five and fifteen. Symptoms usually develop more slowly.

What remains unaffected by MND?

In the majority of cases the intellect and memory are not affected, nor are the senses of sight, hearing, taste, smell and sensation.

The bowels and bladder are not directly affected by the disease. Incontinence is not a usual feature of MND, but constipation can occur, especially when people become less mobile or have to change their diet due to swallowing difficulties.

Is Motor Neurone Disease painful?

The neurons carrying sensory messages of pain, touch, heat, cold and pressure from the skin and deeper tissues back to the spinal cord and brain are unaffected.

However, while most people experience little or no pain at any stage of the illness, others may experience some pain or discomfort.

Examples of the types of pain are:

Cramps in the muscles especially those of the legs. These are most common in the early stages of the disease and may occur with exercise or at rest.

Weakness of the muscles around a joint allows excessive strain to be transmitted directly through the ligaments and soft tissues. This results in pain and stiffness in the joint. The problem most commonly affects the shoulder joint. Care should be taken not to pull on the arms when assisting transfers or to leave a weak arm hanging unsupported.

Inactivity of muscles of the shoulder area may lead to a "frozen shoulder" problem, where movement at the shoulder causes pain.

The loss of normal posture control, secondary to muscle weakness, can result in pain in the muscles of the neck, shoulder, hip and knees. The discomfort is characteristically improved by changing the patient's posture or position. This problem is worse in people who are too weak to readily move or reposition themselves. Carers need to know how and when to readjust the position of patients with MND in order to minimise this problem. Electrically operated beds and chairs can assist the person with MND to improve their mobility and subsequent comfort.

Unpleasant feelings of exaggerated sensitivity of the skin. The cause of this uncommon problem is unknown.

Pain can be treated using a number of physical measures. Special cushions and mattresses provide extra 'padding' especially if patients have lost weight. Prevention of pain arising from stiff shoulders and other joints is possible through gentle passive exercise of those limbs which are too weak to be moved by the patient's own efforts. An example of physical treatment is the use of hot packs - or even a warm bath. Similarly, transcutaneous electric nerve stimulation (TENS) may also be of value. Although TENS machines are expensive, they can be obtained on loan or hired. Information about sources of supply in each State are available from the local Motor Neurone Disease Association. Physical treatments are best accessed through a physiotherapist.

Depending on the symptoms being experienced by the patient, Doctors can prescribe a range of medications for pain.

Are there alternatives to medication for dealing with pain and anxiety?

Alternatives to medication are available.

Relaxation therapy is of great value to all people, including those living with MND especially if they are experiencing breathing or swallowing problems. It is a simple but expedient way to reduce anxiety. A variety of relaxation tapes is available, as is help from support people in learning relaxation techniques.

When considering alternatives, patients are encouraged to check with their treating doctor before embarking on alternative or additional treatment to that being given already.

Some people find acupuncture, as an alternative to or in conjunction with traditional medicine, useful in the treatment of pain. Care must be taken, however, to ensure that treatment is given by a qualified acupuncturist who uses sterilised needles.

Is there a cure?

At present there is no cure, but coordinated research is being carried out across the world and encouraging progress is being made.

Costly and unproven therapies are sometimes recommended by well meaning people. Patients should seek professional advice before embarking on unproven therapies. It is important to discuss the likely benefits of expensive therapy compared with, for example, changes to the home, employment of additional home assistance, or the peace of mind of the patient who wishes to leave his/her family well provided for.

Researchers are developing and trialing drugs, for example to slow down the progression of the disease. For further information you should speak to your Neurologist or contact the Motor Neurone Disease Association in your State.

You may hear through TV or other media of new advances. You should always check with your own Doctor or care team before trying these.

Although there is no cure at this point in time, it is quite untrue that "nothing can be done for you". A great deal can be done to maintain quality of life and address many of the effects of MND.

Health Professionals who may be of assistance.

Associated with support from family and friends, other people who may help include GPs, Neurologists, Occupational Therapists, Physiotherapists, Speech Pathologists, Psychologists, Home Care Nurses and Social Workers. This list is not exhaustive.

Because MND is not a common disease, even experienced General Practitioners may not have seen a case previously. Your General Practitioner can carry out the basic neurological examination, and recognise symptoms which indicate the need

for referral to a Neurologist. They will then liaise with the Neurologist and allied health care providers in order to provide the best possible standard of care and maintain quality of life.

The role of the Neurologist is to confirm the diagnosis and exclude other conditions which may be amenable to different treatments. The Neurologist will be able to monitor the progress of the disease and help in initiating supportive care at appropriate times. In some cases the Neurologist may seek a second opinion from another Neurologist.

An Occupational Therapist can help to maintain mobility, function and independence. They can advise on different ways of performing tasks, and the selection, acquisition and adaptation of disability aids. Occupational therapists can visit private homes to advise on equipment or home alterations.

A Physiotherapist can help to maintain the physical activity and mobility of the body, make the most of muscle strength, and alleviate or prevent joint stiffness and pain caused by wasting, spasticity and cramps. Physiotherapists can instruct carers in a range of passive exercises, and the techniques of positioning a person with MND so as to avoid injury to themselves or to the person for whom they are caring.

A Speech pathologist specialises in the management of communication and swallowing problems. Communication aids range from simple manual systems such as alphabet boards, to voice-synthesising computers that can be operated from a single soft-touch switch. Speech pathologists can assess and advise on different swallowing techniques, food preparation and selection. This may be done in collaboration with a dietitian who will suggest a diet appropriate to the individual's needs.

A Community or District Nurse can provide a range of nursing and ancillary services to people in their own homes. Services are usually obtained by referral from a health professional such as the family doctor. Fees may be charged. Provision of services varies from State to State; check with the Motor Neurone Disease Association in your State.

A Social Worker, psychologist, or other qualified counsellor can provide counselling about coping with the psychological and emotional aspects of MND. In addition a Social Worker can offer advice on the services and benefits (financial, legal, accommodation etc.) available in the local community, or through Local, State or Commonwealth Government.

It is possible that therapists, and even the family doctor, may not have had previous experience of MND. Every patient is different. It is important to check major questions with the treating Neurologist. The Motor Neurone Disease Association in each State and the ACT can provide printed information on the management of MND and the support available in that State.

Some MND Associations can offer support and advice from professional staff and experienced volunteers. These people work together with those who have personal experience of living with MND and their carers, to provide support and advice based

on a wide understanding of the nature of the issues associated with each stage of the disease.

Will I develop mobility problems?

Most people with MND do develop problems related to mobility, to a greater or lesser extent.

It is unlikely that people living with MND will stay in bed for long periods, but they may need help moving around. Some people have a tendency to fall, sometimes without warning.

Occupational therapists and physiotherapists can advise regarding access around your home and assist in the provision of appropriate aids to improve mobility and safety.

Modifications to the home, such as the fitting of hand rails, may be arranged through the Home and Community Care (HACC) program.

Advice about home alterations is also available through the Home Renovation Advisory Program, a service of the Department of Planning & Development in each State.

Will I be more tired than usual?

Fatigue is common with MND. By recognising the factors that worsen symptoms and by learning how to conserve energy, people can greatly improve their quality of life. Some strategies suggested are:

Plan activities in advance.

Take regular rest periods. Rest between activities and before going out.

Don't exercise to the point of excessive fatigue, cramps, or muscular weakness. Discuss with a physiotherapist exercises to alleviate stiffness, muscular tension, or pain.

Pace yourself by moving slowly, with frequent rests. Take a few breaths before recommencing a task. Keep heavier tasks for times of greater energy. Stop if you become breathless.

Use mechanical aids to make tasks easier.

Take shortcuts where possible, and sit rather than stand.

Try to establish a regular sleeping pattern.

Make your environment easy to move around in.

What can I do if I can't sleep well?

You may experience difficulty sleeping because of the emotional distress of receiving a diagnosis of MND, or the stress of living with MND. In these circumstances you are encouraged to speak with your treating Neurologist.

Sleeping difficulties may also be caused by discomfort arising from immobility, pain due to stiffness of joints or muscles, excessive saliva or dry mouth, or a variety of other reasons.

Some strategies suggested are:

Try not to remain in the same position in bed for too long.

Satin or silk sheets may help moving about in bed.

An electric bed can be adjusted to a variety of positions at the push of a button. Side rails, bedside furniture and extra pillows can give added support.

A segmented overlay mattress and specialised pillows distribute body pressure more evenly.

Muscle spasticity or pain from muscle tightness and joint stiffness may be relieved with medication prescribed by your doctor. Non-prescription analgesics such as aspirin or paracetamol relieve mild discomfort.

Joint pain may be relieved by stretching or range-of-motion exercises before retiring. A physiotherapist may be able to find the origin of the problem and recommend a remedy.

Breathing may be helped by elevating the head and chest with an extra pillow or two. This relieves pressure on the diaphragm and improves lung expansion.

Before going to bed, avoid over-exertion, caffeinated drinks, smoking, and heavy meals. Too much food puts pressure on the diaphragm and aggravates breathing problems.

Retire at the same time each night and reduce daytime napping. Make sure the bedroom is quiet, cool, dark, and comfortable. Light bedclothes allow easier movement in bed.

Impaired swallowing reflexes may cause a build up of saliva or mucus. This is annoying, especially for those with an ineffective cough. Sleep with the head elevated to prevent secretions from moving toward the cough reflex centre.

A slow rhythmic back rub or a light massage of aching muscles promotes relaxation. One exercise for reducing stress is deep abdominal breathing. Soft music or reassuring conversation can promote sleep.

Will there be breathing difficulties?

Many people with MND have decreased lung capacity because of muscle weakness. Any activity that requires increased respiratory effort will therefore be harder for a person with MND. Some people find their cough is less forceful than before, making it more difficult to clear their throat. Patients with swallowing difficulties can be more prone to chest infections. People with MND may also experience the

feeling of not being able to get enough air, rather like the sensation of being in an overcrowded room. If shortness of breath becomes distressing, your doctor should be consulted immediately.

Strategies suggested are:

Position is important. Sitting in a slumped position restricts lung capacity. Sitting up may be better than lying down.

When sitting, the bottom should be well back in the chair, the back straight and well supported, the head in the midline and also well supported.

If excessive saliva or mucus is a problem, seek the advice of your doctor and speech pathologist about controlling it. A physiotherapist can also teach assisted coughing.

Avoid contact with people who have colds or flu, and consult your doctor about the need for influenza vaccination.

Try to keep the bedroom and living areas at a comfortable, steady temperature (around 18 and 21 degrees respectively). Keep the rooms well ventilated and avoid smoking or being in the presence of smokers.

Routine chest physiotherapy can be beneficial, but should not be too vigorous. The physiotherapist may also suggest a program of breathing exercises to help maintain lung expansion. Shortness of breath may be helped by breathing in a calm and purposeful way until the sensation has passed.

Your doctor can prescribe medication which can significantly help the symptoms of breathlessness.

Assisted ventilation may help some patients.

Will there be swallowing problems?

In some cases people experience swallowing difficulties which require adaptation of the process of eating and drinking. In conjunction with the advice of a speech pathologist the following strategies have been found to be helpful:

Modify the texture and consistency of food and drink. Minced and vitamised foods are usually easier to swallow. Thickened drinks flow more slowly and are thus easier to control.

Small amounts of food and drink per swallow reduce the risk of inhalation. In most cases drinks should be sipped and each intake of food about a teaspoonful.

Allow more time to eat and drink. Don't try to keep pace with others during meals. Eat and drink privately if you feel pressured in company.

Eat foods you like - taste is a strong stimulus for the swallow reflex. Maintain variety by vitamising foods separately to retain the taste and colour of the food.

Swallowing is generally safer when you are sitting as straight as possible with the head upright. Avoid tilting the head back or bending it forward towards your chest. Some people find swallowing easier if the head is slightly forward, as if sniffing the air.

Find a cup that suits your drinking style. Lightweight cups with a wide, flared top are suitable. It is usually easier to eat from shallow-bowled spoons. More specialised eating utensils, such as spouted cups and syringes are available. An assessment is suggested to ascertain their appropriateness.

Vibration and icing of the muscles of the face and throat can be useful in facilitating swallowing. A speech pathologist can show you these techniques.

The presence of ropy, tenacious secretions in the mouth and pharynx can make swallowing more difficult. Drinking fruit juices, particularly grape juice, and sucking papaya enzyme tablets help to break down the secretions.

An excellent video regarding swallowing difficulties is available from the Motor Neurone Disease Association of Victoria.

If swallowing becomes impossible or too tiring you may choose to have a gastrostomy. This entails having a tube surgically inserted through the upper abdominal wall into the stomach under a light anaesthetic. Food, usually a proprietary liquid feed, is put down the tube.

Having a gastrostomy does not necessarily mean that you cannot eat. If your swallowing is reasonably safe, you may be able to eat small amount of food for pleasure while having most of your intake via the gastrostomy. This approach may reduce fatigue and should be considered by those who experience marked problems in eating or drinking.

If I need aids and equipment where can I get them?

There is a Commonwealth-funded program for the provision of free disability aids to permanently disabled people living at home, who are not eligible for help under other Government services.

Provision of equipment is arranged by paramedical staff such as physiotherapists and occupational therapists, who assess needs and make application for provision of an aid. An occupational therapist or physiotherapist may be available to accompany you to an Independent Living Centre in States where this type of Centre is established, to test and select equipment.

Aids and equipment are also available from other sources, depending on the State in which you live. Please contact the Motor Neurone Disease Association in your State for details of what is available within your State.

What financial assistance is available?

The Department of Social Security provides information about pensions and other Government benefits. Benefits include:

Domiciliary Nursing Care Benefit. This is a benefit paid by the Commonwealth Department of Human Services & Health in recognition of the effort made when people who would otherwise require admission to a nursing home are cared for at home. It is not means tested, does not affect entitlements to pensions or other benefits, and is not currently regarded as taxable income by the Australian Taxation Office. Application forms are available from the Motor Neurone Disease Association in your State.

Carer's Pension is available to a person providing personal care and attention or constant supervision to a person with a severe physical, intellectual or psychiatric disability.

Health care and concession cards. The Department of Social Security issues various health and concession cards. The cards may entitle the holder to a wide range of concessions covering health, transport, household bills and recreation.

Reduced charges and tariffs are available to people who receive Social Security benefits, or who experience financial difficulty. These reductions apply to electricity, gas, telephone rental, council rates and car registration.

Mobility Allowance is available from the Department of Social Security to people who are unable to use public transport and are engaged in employment, training or volunteer work for a minimum of ten hours per week.

Disability Support Pension is available from the Department of Social Security to people who have a physical, intellectual or psychiatric impairment of at least 20% that causes them a continuing inability to work for at least two years.

What support is available for carers?

As most people with MND remain at home, the needs of the whole family, and particularly of the primary care-giver, must be taken into account.

Many carers find that contact with others in the same situation can be a great source of information and support. Support groups, set up by the Motor Neurone Disease Association, exist in most States and the ACT.

In some cases these groups provide an opportunity to talk with professionals and other support staff. However, in all cases, people living with MND and their carers meet together to share experiences, learn from each other, share a few laughs and maybe shed a few tears together.

MOTOR NEURONE DISEASE ASSOCIATIONS

Motor Neurone Disease Association of New South Wales,

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Phone 03 9596 4761 Fax 03 9596 8005

Motor Neurone Disease Association of Queensland,
P O Box 259, Corinda 4075
Phone 07 396 8360

Motor Neurone Disease Association of South Australia,
Neurological Resource Centre
23A King William Road, Unley 5061.
Phone 08 357 8909 Fax 08 357 8876

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Motor Neurone Disease Association of Tasmania,
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Further copies of this booklet can be ordered from the Motor Neurone Disease Association of Australia by phone or fax as above, or by writing to them at Reply Paid 38, P O Box 262, Caulfield South, Vic. 3162.

Acknowledgments

Motor Neurone Disease Assn. of Vic. "What is Motor Neurone Disease?"
"Living with Motor Neurone Disease"
"Maintenance of Independence"

Mulder, Dr. D.W. (ed.) "The Diagnosis & Treatment of Amyotrophic Lateral Sclerosis", Tucson, Arizona, 1979.

Oliver, D. "Motor Neurone Disease". The Royal College of General Practitioners: London. 1994.

The Neurological Centre, Westmead, Australia. "Motor Neurone Disease". (c) 1993 Helke Pty Ltd.