Progressive Muscular Atrophy

This information sheet provides an explanation about a relatively uncommon and sometimes slower progressive form of motor neurone disease (MND), called progressive muscular atrophy (PMA).

It is a rare form of motor neurone disease, and as such comes under the support of the Motor Neurone Disease Association.

What is Progressive Muscular Atrophy?

PMA is a disorder of the lower motor neurones. In comparison, the more typical forms of MND have both upper and lower motor neurone involvement.

Lower motor neurones are a group of nerve cells in the spinal cord or in the brainstem that control muscles of the limbs or body. The lower motor neurone is at the bottom of the chain of command, but damage results in a loss of muscle control, weakness and muscle wasting (atrophy).

In PMA there is gradual loss of the lower motor neurones, so the muscles become steadily weaker and more wasted, causing weakness in the area they serve. This results in progressive muscle weakness, fasciculation’s (rippling effect under the skin), and shrinkage in muscle bulk and weight loss.

PMA affects 5-7% of all people living with MND. Life expectancy, although variable, is generally longer than the average for all cases of MND, averaging 5 – 10 years.

Kennedy’s Disease is a rare form of Spinal Muscular Atrophy, with some features in common with PMA. This can cause confusion during diagnosis.

Please see Information Sheet 2 - Kennedy’s Disease (details on how to access other publications are given in the section ‘Further information’ at the end of this sheet).
In the diagram to the right, the red or dark line indicates upper motor neurones, which are NOT affected by PMA.

The green or light lines are the lower motor neurones where changes occur in PMA.

PMA is held to be a form of MND and, especially in the first 4 years after diagnosis, clinical evidence of upper motor involvement may become apparent in some people. In others, the physical effects may remain those of fairly pure lower motor neurone damage.

PMA, like other forms MND, occurs sporadically in the majority of affected individuals. Sporadic is the term used for cases of the disease where there is no family history.

People with inherited or so-called familial MND (the term used when more than one member of the family has been diagnosed with MND), can present with the PMA form of MND.

How is it diagnosed?

PMA, like other forms of MND, can be difficult to diagnose. It has to be recognised that although the majority of people with PMA actually have the ‘lower motor neurone’ form of MND, about one third may have other forms of lower motor neurone disorders. It is vital for your doctor or neurologist to review the diagnosis from time to time, as this helps to identify other lower motor neurone disorders, eg Kennedy’s disease, spinal muscular atrophy, multifocal motor neuropathy or occasional autoimmune nerve damage.

The diagnosis is clinical and usually based on observation, supported by additional investigations such as EMG (electromyography) and the exclusion of other possibilities, such as spinal cord injury, tumours and motor neuropathies as mentioned above from other causes.

What are the tests?

The tests which might be performed include:

- MRI scans to exclude abnormalities of the brain or spinal cord
• Nerve conduction studies (small electric shocks to check speed of nerve conduction to the muscles)

• Electromyography (EMG) where small needles are placed in individual muscles to check for any dysfunction (abnormal in lower motor neurone disease)

• Blood tests to check for other possible diseases

Electrophysiological tests (so called central motor conduction studies or magnetic stimulation) on the brain may also be carried out in an attempt to identify any upper motor neurone component which may not be evident on clinical examination.

An EMG test is a good way of detecting lower motor neurone disease, but has to be put into context with the results of other tests carried out, to eliminate all other possible diseases.

What are the symptoms?

PMA is characterised by:

• Muscle weakness
• Muscle wasting
• Fatigue
• Fasciculations
• Cramps
• Muscle twitching
• Loss of reflexes.

PMA starts most frequently with weakness in one hand/arm, although it can be in one foot/leg and very rarely the tongue. Weakness spreads to other muscles as the disease progresses. Cramps usually precede weakness and muscle twitching also occurs at an early stage. Both cramp and muscle twitching occur in normal healthy individuals and therefore by themselves do not indicate any cause for concern.

Not having upper motor neurone involvement means that people living with PMA do not have the stiffness (spasticity) in their muscles, nor the very brisk reflexes and the uncontrollable emotions seen in other forms of MND.
How might I be affected?

The first noticeable effects depend on where the disease starts.

If the initial presentation is in the legs, then stumbling or difficulty climbing stairs may be noticed. If the initial weakness occurs in the arms, then loss of dexterity or an increase in dropping things may be the first signs. Cramps, twitching in muscles, aching muscles and general fatigue, can precede obvious weakness.

For a tiny percentage, PMA doesn't spread from the initial area affected and although disabled, such individuals have a relatively benign course to their disease. An example could be bi-brachial amyotrophy (the so called ‘flail arm syndrome’) which affects the arms, but often nowhere else for a prolonged period. However, it must be remembered that progression of PMA can be very variable.

For the majority, PMA continues to spread causing increasing disability and speech and swallowing can also become difficult. As breathing muscles are controlled by lower motor neurones, they can also be affected.

The final pathway for PMA is much the same as for other forms of MND, but without major upper motor neurone involvement and usually at a slower rate of disability progression.

However, as with all MND, PMA follows a different time course in each person.

Management of PMA

As with other forms of MND, the disease is managed by support of the individual’s disabilities:

- Assistive equipment may be needed for leg and arm weakness.
- Communication aids may be required.
- Feeding may become difficult and require assistance, e.g. via a PEG tube.
- Breathing may require support via non-invasive ventilation (NIV).

Various information sheets are available from the MND Association regarding NIV (please see ‘Further information’ at the end of this sheet for details on how to access other publications).

In all areas of support for the disease, professional assessment is advised, so that care is tailored to the individual.
Email Based Support Group

PMA is generally a longer term condition, so you may find it beneficial to have some contact with others in a similar situation. The MND Association has developed an email based support group for people living with rarer forms of MND (please see the end of this sheet for details on how to access further information).

Other organisations

The following organizations are included to give you an idea of where to look for further information on MND and PLS. Please note that the MND Association does not necessarily endorse all of the information they supply.

World Federation of Neurology ALS Page
A research group uniting people and studies on MND across the world.

World Federation of Neurology
National Neuroscience Centre
Beaumont Hospital, Dublin, Ireland
Trinity College Dublin
Telephone: 353 1 8376528
E-mail: orla@hardiman.net
Website: www.wfnals.org/index.html

The ALS Association
A leading American society working towards a cure for ALS and all types of MND and compassionate care for those living with the disease.

The ALS Association
National Office, 1275 K Street NW, Suite 1050, Washington, DC 20005, USA
Email: through the website contact page
Website: www.alsa.org

Neurology
An online neurology journal featuring latest news on a variety of neurological conditions, research and studies.

Website: www.neurology.org

References

Kim, W.-K. et al. Study of 962 patients indicates progressive muscular atrophy is a form of ALS. Neurology 73, 1686–1692 (2009)
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Further information

If you would like to take part in our email based support group for people living with rarer forms of MND, please visit the following link on our website: www.mndassociation.org/PMA-PLS

You may also find the following MND Association publications helpful:

- What is motor neurone disease (MND)?
- Symptoms
- Information Sheet 11 - PEG Feeding - making the decision
- Information Sheet 14A – Understanding how motor neurone disease (MND) might affect breathing
- Information Sheet 14B – Ventilation in motor neurone disease (MND)
- Information Sheet 14C – NICE guidelines on non-invasive ventilation (NIV)
- Information Sheet 14D – Troubleshooting when using non-invasive ventilation (NIV)
- Information Sheet 14E – Air Travel and non-invasive ventilation (NIV)

If you have any questions about the information on this sheet, please contact the MND Connect team.

Downloads of all our information sheets and most of our publications are available from our website. You can also order our publications direct from the MND Connect team, who will also be able to advise on individual needs:

MND Connect
MND Association, PO Box 246, Northampton NN1 2PR
Telephone: 08457 626262
Fax: (01604) 638289
Email: mndconnect@mndassociation.org

MND Association website and online forum
Website: www.mndassociation.org
Online forum: http://forum.mndassociation.org/ or through the website
We welcome your views

Your feedback is really important to us, as it helps improve our information for the benefit of people living with MND and those who care for them.

If you would like to provide feedback on any of our information sheets, you can access an online form at: www.surveymonkey.com/s/infosheets_1-25
Or request a paper version by email: infofeedback@mndassociation.org

Or write to:
Information feedback
MND Association
PO Box 246
Northampton NN1 2PR