

# The Diagnostic Dilemma of Progressive Muscular Atrophy

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## ABSTRACT

Progressive muscle atrophy is a rare subtype of motor neuron disease that affects only the lower motor neurons and presents as asymmetrical rapidly progressive muscle weakness, atrophy and normal sensations. The diagnostic electrophysiological findings are denervation potentials in three out of four body segments (bulbar, cervical, thoracic and lumbosacral). The disease is fatal and the management is supportive. We present the report of a 45-year-old female patient who presented with unilateral foot drop and rapidly progressed to profound weakness in muscles of all limbs, neck and back along with dysarthria and dysphagia. She had been operated twice for suspected cervical and lumbosacral intervertebral disc herniations and ultimately guided in right direction after muscle biopsy, nerve conduction studies and electromyography.

**Key Words:** *Progressive muscle atrophy. Motor neuron disease. Nerve conduction studies. Electromyography. Magnetic resonance imaging. Unilateral foot drop.*

## INTRODUCTION

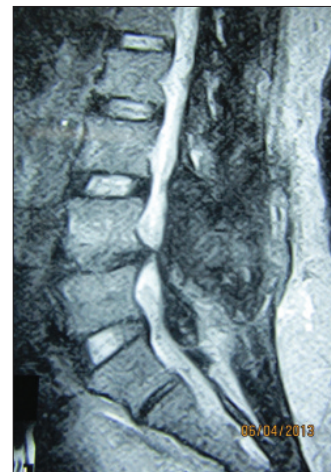
Progressive Muscle Atrophy (PMA) is a rare subtype of Motor Neuron Disease (MND) which affects only the lower motor neurons. The diagnosis is based on progressive extremity weakness and atrophy with a rapid evolution and characteristic findings on Nerve Conduction Studies and Electromyography (NCS/EMG).<sup>1</sup> The disease may be confused with polyradiculopathy and if Magnetic Resonance Imaging (MRI) is supportive, the patient may inadvertently be exposed to surgical procedures to remove root compression.

We present here a case of PMA who developed progressive ascending weakness starting with unilateral foot drop and underwent spinal surgery twice for lumbar and cervical radiculopathies.

## CASE REPORT

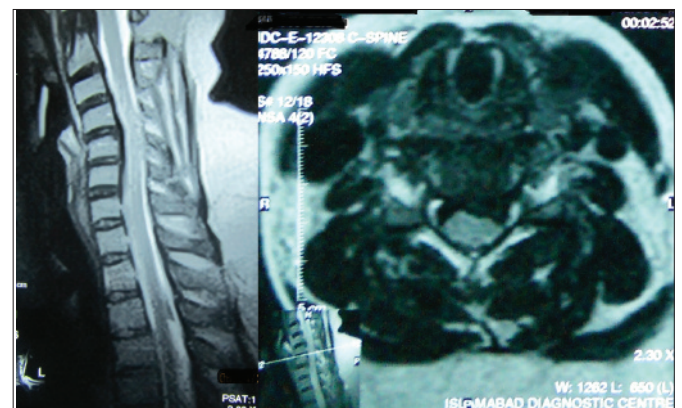
A 45-year-old female presented with complaints of loss of power in all limbs, difficulty in speech and swallowing and dependence in all activities of daily living for the past 3 years. At the outset, she progressively developed foot drop on left side causing her difficulty in walking and pain in left leg. She consulted a neurosurgeon who advised her an MRI of the lumbosacral spine. Laminectomy based on left postero-lateral bulge of L<sub>4</sub>/L<sub>5</sub> intervertebral disc (Figure 1) was performed. There was no improvement and her problem progressed. During the next 2 years, she developed weakness in her right leg accompanied by back pain and became completely

bed ridden. She did not have any sensory loss. A request for NCS/EMG of lower limbs discovered lumbosacral polyradiculopathy involving L<sub>4</sub>, L<sub>5</sub> nerve roots bilaterally and L<sub>3</sub>, S<sub>1</sub> roots on left side. One year



**Figure 1:** Sagittal film of MRI scan of lumbosacral spine showing left postero-lateral bulge of L<sub>4</sub>/L<sub>5</sub> intervertebral disc.

back, the patient also started noticing weakness in her left hand. MRI of the cervical spine showed degenerative disc disease with associated disc osteophyte complexes at C<sub>4</sub>/C<sub>5</sub> and C<sub>5</sub>/C<sub>6</sub> levels causing left foraminal stenosis at C<sub>5</sub>/C<sub>6</sub> level (Figure 2). MRI of thoracic spine on same date and MRI of brain were unremarkable. She was operated in the region of cervical spine but she developed in neck an additional pain and loss of power in her right arm.



**Figure 2:** Sagittal and axial films of MRI scan of cervical spine showing degenerative disc disease with associated disc osteophyte complexes at C<sub>4</sub>/C<sub>5</sub> and C<sub>5</sub>/C<sub>6</sub> levels causing left foraminal stenosis at C<sub>5</sub>/C<sub>6</sub> level.

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For the past 6 months, the patient had been facing difficulty in swallowing and slurring of speech. She had also developed weakness in her neck and was unable to hold her neck. She was having a disturbed sleep and episodes of low mood and excessive weeping. She was completely dependent in self-care and mobility, however, remaining continent for bladder and bowel.

On examination, she was bed-ridden and had an average build with stable vital signs. She was well-oriented but had dysarthria. The passive ranges of motion of cervical spine, shoulder and hip joints were full. Both upper and lower limbs were flaccid. She had a power grade of 0/5 in all key muscles of the four limbs. All sensations were intact. Deep tendon reflexes were absent in all limbs and plantars were equivocal. Rest of the systemic examination was unremarkable.

Her routine laboratory investigations and muscle enzymes were normal. The NCS showed reduced motor amplitudes and normal sensory studies. EMG revealed denervation potentials with no voluntary activity in tibialis anterior, extensor hallucis longus, rectus femoris, first dorsal interosseous, extensor digitorum communis, pronator teres and thoracic paraspinals on both sides placing her in El-Eschorial electrodiagnostic criteria,<sup>2</sup> for diagnosis of Amyotrophic Lateral Sclerosis (ALS) and its variants (like PMA). Muscle biopsy of the affected muscles turned out to be normal.

The patient's attendants were briefed about the disease, its outcome and trained in nursing care, feeding techniques and therapeutic exercises. The patient was given analgesics, mood stabilizers and psychotherapy sessions. She refused gastrostomy tube placement and was taken home by her attendants.

## DISCUSSION

First reported in 1850,<sup>3</sup> PMA comprises of 4% of all MND cases.<sup>4</sup> It can occur sporadically (in the majority) or in familial forms and can affect one or all body regions. It characteristically involves lower motor neurons which differentiates it from ALS and other variants of MND. The disease commonly manifests as asymmetric limb weakness, muscle atrophy and fasciculations gradually progressing to other limbs. Maintaining erect posture and holding neck becomes difficult owing to paraspinals' inefficiency. Diaphragm weakness produces orthopnea and sleep disturbance. Bulbar muscles are affected at

the last if at all. All sensations remain intact. There is usually no cognitive dysfunction.

PMA is a diagnosis of exclusion. Polyradiculopathy, spinal muscular atrophy, myopathy, multifocal motor neuropathy, inclusion body myositis, and chronic inflammatory demyelinating polyneuropathy have to be ruled out first.<sup>5</sup> Patients may be mistakenly treated conservatively or surgically for other common likelihoods. Useful investigations are muscle enzymes, muscle biopsy, NCS/EMG and MRI. Presence of denervation potentials in three out of four body segments (bulbar, cervical, thoracic and lumbosacral) with normal muscle enzymes and muscle biopsy is usually conclusive of PMA.<sup>5</sup>

The disease is always fatal. The course is relentlessly progressive but prognosis is better than classical ALS.<sup>6</sup> Management consists of a drug-trial of Riluzole, nursing care, respiratory therapy, adequate nutrition, sufficient analgesia and psychotherapy.<sup>5</sup> This case presented with asymmetrical progressive weakness and was mistakenly operated twice owing to diagnostic confusion. After all, NCS/EMG came up to the patient's rescue and the management was guided in right direction.

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