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A Challenging Rare Motor Neuron Disease to Diagnose

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ABSTRACT

Mills hemiplegic variant is a progressive motor neuron disease characterized by spastic ascending or descending hemiparesis or hemiplegia with no sensory involvement. We presented a 53years old male patient with a history of present complaints was difficulty on walking, and difficulty in speech then he developed left lower limb weakness followed by left upper limb weakness for the last 6 months. Electromyography showed a neurogenic pattern with denervation with reinnervation. Then, he successfully accomplished his treatment. The biggest challenge in making a diagnosis is ruling out other conditions that may have symptoms similar to this disease entity.

Keywords: Mills hemiplegic, Electromyography, Motor neuron disease

INTRODUCTION

MND is a progressive disorder in which degeneration of upper and lower motor neurons leads to progressive weakness of bulbar, limb, thoracic and abdominal muscles with relative sparing of oculomotor muscles and sphincter function. Typically, death from ventilatory failure occurs within five years of commencement. Most cases of sporadic (90-95%), whereas 5-10% are familial, usually with the autosomal dominant transmission. Many other motor system and anterior horn cell disorders are recognized. An unusual form of MND occurs in the Western pacific associated with parkinsonism and dementia.[1]

Mills' condition, which includes progressive, ascending, or descending hemiplegia with no major sensory impairment, was initially reported by Mills in 1900. In contrast to being a form of primary lateral sclerosis, this disease was acknowledged as unilateral corticospinal tract degeneration. The syndrome's nosological status is now unclear because it was only ever defined based on clinical assessment.[2]

The anterior horn cell is a motor neuron that extends from the anterior portion of the grey matter in the spinal cord to the skeletal muscle. The degeneration of these cells results in muscular denervation and weakening, which is a defining aspect of this illness. The weakness affects mostly the proximal muscle of the lower extremities which can vary in severity and age of onset.[3]

$CD4^+$ T cells are commonly divided into regulatory T (Treg) cells and conventional T helper (Th) cells. By activating other effector immune cells, Th cells regulate adaptive immunity against infections and cancer. Treg cells are CD4+ T cells that are responsible for regulating potentially harmful Th cell activity.[4]

Twenty-six cases of Mills' syndrome have been reported since 1906, mostly as single reports, but only 16 of these conformed to Mills' original description of an idiopathic, isolated, progressive, spastic hemiparesis. Of the remainder, two were diagnosed with ALS and a further two

with multiple sclerosis one evolved into PLS and also developed an extrapyramidal syndrome, one was attributed to lacunar infarcts and two were ascribed to frontotemporal dementia/motor neuron disease. In two further cases, the presentation involved significant cognitive or neuropsychiatric abnormalities.[5] Advances in the diagnostic laboratory, electromyography (EMG), and neuroimaging testing verified the image of mills syndrome's gradual unilateral ascending weakness accompanied by pyramidal dysfunction.[6]

CASE REPORT

A 53-year-old male with complaints of difficulty on walking and difficulty in speech. He was normal for the previous two years, but now he has left lower limb weakness followed by left upper limb weakness. Hypertension and diabetes mellitus were both recognized conditions.

Examining the MRI data revealed potential neurodegenerative disorders, grade 1 chronic small artery ischemia alterations, and a negative autoimmune panel. On his 1st EMG which showed no active or chronic degenerative changes and motor neuron disease. Later, he had ayurvedic treatment, but it restricted his mobility and made his ailments worse. Finally, on his 2nd EMG report, it was found that the anterior horn cell participation in the S/O neurogenic pattern with denervation and reinnervation alterations (Table shows EMG report). T and B markers by flow cytometry report showed that IMP- T regulatory cells constitute 6.25% of VD4+T cells was normal.

	Left ABP, deltoid	**LT. Vastus lateralis**	**Left biceps**	**Rt deltoid**
Spontaneous	Fibrillation/ fasciculation positive sharp	fasciculation	fibrillation	Nil
MUAP				
Amplitude	Prolonged	-	Prolonged	-
Duration	large		large	-
Polyphasia	+ +	+	+	-
Stability	unstable	unstable	unstable	unstable
Recruitment				
Interference	incomplete (poor effort)	Incomplete (due to poor effort)	complete	Unable to activate

Table 1: EMG Report

He has done a blood investigation which showed HBA1C 8.8 was under the treatment of inj. actrapid according to scale, creatinine level was 0.70, liver function test and complete blood count was normal. ECHO reported LVEF 60%, mild LV dysfunction, and Trivial MR/TR. PASP=30mmhg. Other neurological examinations accounted for were normal.

He had sent his sample for a Paraneoplastic panel which was positive for an unclassified antibody. Hence with a possible hypothesis of secondary MND under IVIG 30gms for 5 days, a total of 150grms with premedications inj. Dexa 4mg for every cycle. T. Sizodon 0.5mg for anxiety disorder. He had a significant restrictive disease, as evidenced by the PFT data, and required pulmonary rehabilitation. He is also under the treatment of antihypertensive, antiplatelet, antipsychotics, and other supportive medications. Physiotherapy and swallow therapy opinion was given on a regular basis. He became improved neurologically and hemodynamically stable.

DISCUSSION

In the last ten years, genetic understanding of motor neuron disease (MND) has advanced significantly, with the recent discovery of a connection between MND and mutations in TBK1 and NEK. Age and site of onset, rate of disease progression, and clinical syndrome differ significantly between cases, presenting difficulties for diagnosis and disease management. In Scotland, there is a predicted annual crude incidence of MND of 2.38 per 100,000 of the population. Mean survival from symptom onset to death is 2.8 years, albeit with a variable trajectory depending on the clinical syndrome.[7]

Motor neuron disease occurs on a spectrum, with varying degrees of involvement of upper and lower motor neurons. MNDs are distinguished from other neuropathies or neuronopathies by the pattern of motor and/or sensory involvement. Confirming the diagnosis may initially be difficult until the full clinical features are manifest. There is a significant differential diagnosis to consider for all types of the disease, including curable disorders, and hence professional neurological advice should always be sought.[8] In contrast to MMN, antigenic targets in CIDP remain largely elusive, although IgG4 antibodies against the paranodal proteins neurofascin-155 and contactin-1 have recently been described and are associated with a severe sensory and motor CIDP phenotype with poor response to IVIg, favourable response to rituximab.[9]

CONCLUSION

All forms of motor neuron disease spectrum (ALS, PLS, or UMN-ALS) that manifest as hemiplegia or as an asymmetrical pattern of involvement can be classified as Mills syndrome. A syndrome is a grouping of symptoms and signs that may be caused by a variety of factors. In this case, it was challenging to diagnose because the symptoms were similar to those of other neurological illnesses. The patient was diagnosed and successfully completed IVIG treatment.

Declaration by Authors

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