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Case Report

Multiple System Atrophy in a Filipino 36-year-old Female Presenting as Progressive Gait Ataxia - A Case Report

Meleza Mea B Cambalon and Luis C Lagarde*

Graduate of Neurology in Philippines

***Corresponding Author:** Luis C Lagarde, Graduate of Neurology in Philippines.

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Abstract

Multiple system atrophy (MSA) is a rare, progressive, fatal, neurodegenerative disorder with incidence and prevalence of 0.6 to 0.7 cases per 100,000 and 3.4 to 4.9 cases per 100,000 population, respectively [1]. This is a case of a 36-year-old woman presenting with predominant features of gait ataxia. On imaging, there was marked cerebro-cerebellar atrophy more prominent in the cerebellum.

Keywords: Multiple system atrophy (MSA); Covid-19; Hypertension

Case

This a case of a 36-year-old woman, Filipino, from Quezon City, Philippines presenting with four-year history of recurring fall episodes and wide-based gait ataxia. She also presented with ataxic dysarthria, occasional dyspneic episodes, dysphagia for both solid and liquid foods, and urinary incontinence. She had multiple admissions and was treated for aspiration pneumonia, covid-19 infection, hypertension, UTI, bronchial asthma and recently pulmonary tuberculosis.

In the subsequent years, she was put on nasogastric feeding due to dysphagia with multiple aspiration episodes. Gait ataxia worsened. She eventually became wheelchair-bound. She did not report any febrile episodes, no headache, and no vomiting during the course of her illness. She had no behavioral changes, however she reported insomnia with frequent awakenings in between sleep.

On physical examination, blood pressure was 120/70 mmHg supine; 90/60 mmHg on standing with support, heart rate of 78 bpm, respiratory rate of 18 cpm, temperature of 36.8 and oxygen saturation of 98% at room air. On neurologic examination, she had moderate cognitive impairment with Montreal Cognitive Assessment - Philippines (MOCA-P) score of 18/30. Cranial nerve, motor, and sensory examination were unremarkable. On cerebellar examination, there was bilateral dysmetria, dysdiadochokinesia, inability to stand without support, and presence of scanning speech. She also had resting tremors on both upper extremities, cogwheel rigidity, hyperreflexia, and presence of babinski bilaterally. Vibration and proprioception sense were intact. There was absence of meningeal signs.

Cranial MRI T1 (Figure 1A) and T2 (Figure 1B) imaging showed marked cerebellar atrophy with shrunken cerebellar follae. Sagittal flair imaging showed widened cortical sulci, fissures and ventricles and markedly shrunken cerebellar follae indicative of marked cerebral-cerebellar atrophy, brainstem was also shrunken (Figure 2A-2B) (Figure 3A-3B).

Figure 1: A-B. T1 (1A) and T2 (1B) axial view showed marked markedly shrunken cerebellar follae indicative of marked cerebellar atrophy.

Figure 2: A-B. Sagittal flair showed marked volume loss as evident on the widening of the cortical sulci, fissures and ventricles and markedly shrunken cerebellar follae indicative of marked cerebral-cerebellar atrophy.

Figure 3: A-B. DWI showed shrunken cerebellar follae and pons (3B) indicative of pontocerebellar atrophy.

Discussion

Multiple system atrophy is a very rare neurodegenerative disease with incidence and prevalence of 0.6 to 0.7 cases per 100,000 and 3.4 to 4.9 cases per 100,000 population [1]. It is the most rapidly progressive of the synucleinopathies which is a group of disorders characterized by the abnormal deposition of the protein α-synuclein (αSyn) in the central and peripheral autonomic nervous system [2]. MSA is sub-classified into a parkinsonian (MSA-P) and a cerebellar phenotype (MSA-C) [3]. The age at onset, prevalence of cardiovascular autonomic dysfunction, sleep disorders, and retinal abnormalities are similar in both phenotypes [4,5]. Patients with MSA have a mean age at onset of 55-60 years, and an average survival from the onset of motor symptoms of 8-9 years, although some pathology-proven cases survived more than 15 years [6,7]. Our patient had her first symptom- onset at a relatively younger age appearing during her third decade of life. Her symptoms predominantly presented with progressive gait ataxia and subclassified as cerebellar phenotype (MSA-C). She initially had frequent fall episodes with wide based gait, which worsened, until she became wheelchair bound.

She also presented with resting tremors, insomnia, and urinary incontinence. Ophthalmologic findings were unremarkable.

Multiple system atrophy (MSA) is difficult to distinguish clinically from other disorders, particularly in the early stages of the disease. An autonomic-only presentation can be indistinguishable from pure autonomic failure. Patients presenting with the cerebellar phenotype of MSA can mimic other adult-onset ataxias due to alcohol, chemotherapeutic agents, lead, lithium, and toluene, or vitamin E deficiency, paraneoplastic, autoimmune, or genetic ataxias [8]. Our patient had no vices and was not diagnosed with any autoimmune, paraneoplastic or malignant diseases. On neurologic examination, she presented with bilateral babinski which is an abnormal upward posturing of the big toe, called as “striatal toe” that resembles a spontaneous extensor plantar response without the fanning of the toes [8].

Cranial MRI is the gold standard imaging technique for the evaluation of MSA presenting with parkinsonian and cerebellar syndromes. A cranial MRI including standard sequences and diffusion weighted imaging (DWI) should be included in the initial evaluation of every patient with suspected MSA [8]. Our patient’s cranial MRI showed marked volume loss as evident on the widening of the cortical sulci, fissures and ventricles and markedly shrunken cerebellar follae indicative of marked cerebral-cerebellar atrophy (see figure 1, 2, 3). The cerebellar atrophy is due to the olivopontocerebellar atrophy degeneration and, to a lesser extent, striatonigral degeneration [9]. The definitive diagnosis of MSA is the presence of pathologically confirmed presence of glial cytoplasmic inclusions consisting of misfolded α-synuclein, a neuropathological hallmark of MSA [10]. Criteria for probable MSA includes sporadic, progressive disease in adults (onset after 30 years old) characterized by autonomic failure, including urinary incontinence, or an orthostatic decrease in blood pressure by at least 30 mm Hg systolic or 15

mm Hg diastolic within 3 min of standing, plus one of the following: Parkinsonism (slowness of movements, rigidity, and tendency to fall) with poor response to levodopa (parkinsonian subtype [MSA-P]) or cerebellar syndrome (wide-based gait, uncoordinated limb movements, action tremor, and nystagmus) (cerebellar subtype [MSA-C]) [11]. Our patient presented with predominantly progressive widebased gait, ataxic dysarthria, and tremors which are manifestations of MSA-cerebellar type. She did not have clinically significant neuropathy, no hallucinations, no family history of ataxia or parkinsonism, and no white-matter lesions which could be suggestive of other neurologic diseases such as multiple sclerosis.

Conclusion

The diagnosis of probable or possible MSA is based on the clinical history and the neurological examination; and the confirmatory definitive pathologic test can be done postmortem. Ancillary tests like autonomic testing, ophthalmologic test, and neuroimaging can assist in the diagnosis and should be performed. Despite the availability of consensus criteria for clinical diagnosis and recent advances in diagnostic techniques, the diagnostic accuracy of MSA remains sub-optimal [8]. Presently, the patient and her family were appraised for the lifelong neurodegenerative course of her illness and the unavailability of definitive treatment. Symptomatic treatment, comprehensive rehab therapy especially for balance and gastrostomy tube insertion were offered.

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