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Case Report

A challenging case presentation of multiple system atrophy cerebellar type: A rare case report from Somalia ☆,☆☆

Nor Osman Sidow, MD[a,c,*], *Abdiwahid Ahmed Ibrahim, MD*[a], *Ismail Gedi Ibrahim, MD*[b], *Mohamed Sheikh Hassan, MD*[a], *Said Abdi Mohamed, MD*[a]

[a] *Department of Neurology, Mogadishu-Somalia Turkey Recep Tayyip Erdoğan Training and Research Hospital, Mogadishu, Somalia*
[b] *Department of Radiology, Mogadishu-Somalia Turkey Recep Tayyip Erdoğan Training and Research Hospital, Mogadishu, Somalia*
[c] *Faculty of Medicine and Surgery, Jazeera Univesity, Mogadishu, Somalia*

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ABSTRACT

Multiple system atrophy is a rare and quickly progressing neurological condition characterized by autonomic failure, parkinsonism, or cerebellar ataxia. It is classified into two subtypes: MSA with predominant parkinsonism (MSA-P) and MSA with predominant cerebellar ataxia (MSA-C). We are presenting here a 54-year-old male with parkinsonism, ataxia, and dysarthria. He was diagnosed with parkinson disease and was given a maximum dose of levodopa but has not responded. After a close neurological evaluation with magnetic resonance imaging of the brain, which shows atrophy of the cerebellum and a brainstem with a hot cross bun sign of the pons, suggestive of multiple system atrophy, he was diagnosed with multiple system atrophy cerebellar type, which is the first time to have this diagnosis in Somalia, which is a low-resource country.

Introduction

Multiple System Atrophy (MSA) is a rare neurodegenerative disorder characterized by autonomic failure, parkinsonism, cerebellar ataxia, and pyramidal tract features [1]. It is referred to as an atypical parkinsonian disorder or Parkinson plus syndrome (PPS), which includes progressive supranuclear palsy (PSP), corticobasal degeneration (CBD), and dementia with Lewy bodies (DLB) [2]. The mean age of onset is 54 years, with a survival rate ranging from 7 to 9 years, which is younger than that seen in parkinson's disease (PD) [3].

Based on the motor manifestations of MSA, it is classified into two subtypes: MSA with predominant parkinsonism

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* Corresponding author.
E-mail address: ziidoow113@gmail.com (N.O. Sidow).

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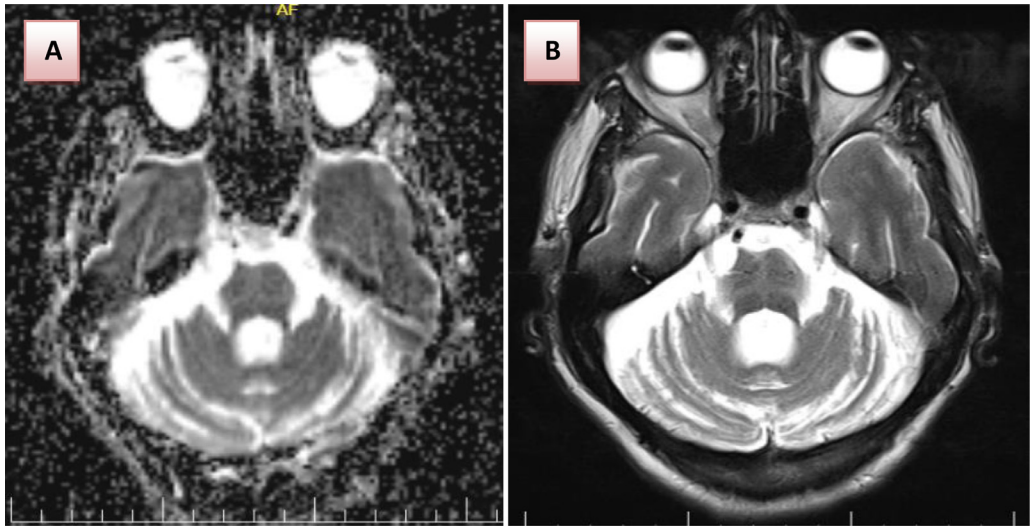

Fig. 1 – Apparent diffusion coefficient (ADC) and T2 sequence of magnetic resonance imaging shows atrophy of the pons and cerebellum (A, B).

(MSA-P) and MSA with predominant cerebellar atakxia (MSA-C) [4]. The prevalence of MSA is 3,4 to 4,9 per 100,000 case populations [5]. The two subtypes have the classic neuropathologic α-synuclein glial cytoplasmic inclusions associated with widespread neurodegeneration in the striatonigral (predominant in multiple system atrophy parkinsonism; MSA-P) and olivopontocerebellar (predominant in multiple system atrophy cerebellar; MSA-C) structures of the brain [6]. The core clinical features of MSA are a varying combination of clinical manifestations that include parkinsonism (bradykinesia plus rigidity and/or tremor), autonomic features, and cerebellar ataxia with pyramidal signs [7]. A definite diagnosis of MSA needs autopsy confirmation of widespread neurodegeneration of α-synuclein inclusions [8]. The clinical diagnosis of MSA-C is based on features and magnetic resonance imaging (MRI) of the brain that show classic hallmark atrophy of the cerebellum and pons (hot cross bun sign) [9,10]. We are presenting here a 53-year-old male who presented action tremor and ataksia associated with dysarthria. After a neurological evaluation with imaging of the MRI brain, showed multiple system atrophy cerebellar type (MSA-C).

Case report

A 54-year-old male presented with kinetic tremor of both arms and dysarthria for one year and a half. The symptoms are progressively increasing. There are associated fasciculations, gait, and limb ataxia. There is no previous history of chronic diseases like diabetes, hypertension, thyroid diseases, stroke, or any other neurological disorders. A neurological examination revealed normal cognition for a mini-mental state examination score of 27. Along with dysarthria, there is limb ataxia with irregular jerky action tremors of both arms. The extensor plantar response was positive. Ocular manifestations were bilateral gaze-evoked nystagmus. No cranial nerve palsy was reported. No dysphagia presented by the patient. There is hypophonic monotony in cerebellar scanning dysarthria seen, which is typically for the cerebellar type. Autonomic features reported for urine urgency and voiding difficulty. No nocturnal aneresis. There is constipation reported by the patient, due to a divorced marital status; erectile dysfunction was denied. There is chronic insomnia complained by the patient.

Previously, the patient went to other clinics diagnosed with Parkinson's disease and was given levodopa at different doses (600 mg) but did not respond. Routine blood investigations, including full blood count, biochemistry, and CSF analysis, show normal results. The electroencephalogram was unremarkable. The autoimmune panel, paraneoplastic investigations, and genetic tests were not performed due to a lack of availability in out hospital and throughout the country. MRI of the brain with intravenous contrast shows Atrophy of cerebellum, and brainstem (Fig. 1A and B), with hot cross bun sign of the pons, suggestive of Multiple System Atrophy/Olivopontocerebellar Atrophy (Fig. 2A and B). So, based on the clinical features and the classic features of the MRI, without responding to the maximum dose of levodopa, the patient was diagnosed with Multiple System Atrophy Cerebellar Type (MSA-C). The patient and the family were explained the diagnosis and the prognosis; no medications were prescribed. For our knowledge, this is the first case reported from Somalia, which is part of a low-resource country.

Discussion

Similar to Parkinson's diseases, 20-75% of cases of multiple system atrophy have a prodromal premotor phase that includes, months to years before the first motor symptoms manifest, sexual dysfunction, bladder incontinence or urgency, orthostatic hypotension, inspiratory stridor, and rapid eye movement sleep behavior disorder [11]. Multiple system atrophy is a rare and quickly progressing neurological condition characterized by autonomic failure, parkinsonism, or cerebellar

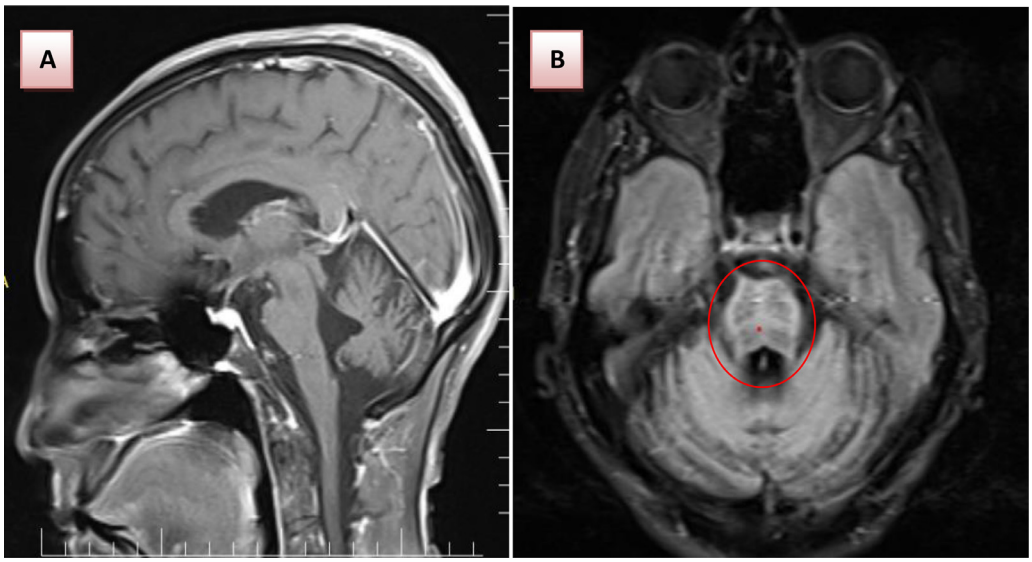

Fig. 2 – Sagittal and fluid-attenuated inversion recovery sequence shows classic atrophy of pons of the H-shaped known as the hot cross bun sign (A, B).

ataxia [12]. Over the last five years, significant progress has been made in understanding the etiology of the disease, new research confirms the pathogenic function of α -synuclein in MSA, shedding light on its epidemiology and genetics [13]. The term MSA was coined in 1969 to refer to three previously reported neurological entities: olivopontocerebellar atrophy (OPCA), Shy-Drager syndrome, and striatonigral degeneration [14]. Based on the prevailing clinical phenotype, the illness is subdivided into a parkinsonian variant (MSA-P) linked with SND, a cerebellar (MSA-C) variant with OPCA and prominent cerebellar characteristics, and a combination of both types, knows as "mixed" MSA [15].

The etiology of MSA is unknown, however, as with other neurodegenerative disorders, a complex interplay of hereditary and environmental processes appears likely [16], Unlike Parkinson's diseases, no one gene mutation is connected to family variants, and no definitive risk factors have been discovered. A loss of function mutation in the COQ2 gene encoding the enzyme that synthesized coenzymes Q10 (COQ10) (4-hydroxybenzoate-polyprenyl transferase), was described in Japanese family and sporadic cases [17]. Parkinsonism, with rigidity, bradykinesia, postural instability, limbic gait, and tendency to fall, characterized the poorly levodopa-responsive motor manifestations of MSA-P [18]. The motor features are rarely asymmetrical resting tremor, whereas irregular postural and action tremor may occur, cerebellar ataxia, wide-based gait, uncoordinated limb movements, action tremor, gazed downbeat nystagmus predominate in MSA-C. Hyperreflexia and a Babinski sign may occur in 0-50% of patients [19]. While there are currently no effective disease modifying medicines for MSA, but parkinsonism or autonomic dysfunction, symptomatic therapies are available [20]. Our case was a 54-year-old male with action tremor, ataxia, and dysarthria for one year and a half, with rapidly progressing clinical manifestations. The patient was diagnosed with Parkinson's disease and was given levodopa for one year with a maximum dose of 600 mg per day, but did not respond to the motor symptoms. after clinical and neurological evaluation with an MRI brain of classic features diagnosed as multiple system atrophy cerebellar type (MSA-C). Unfortunately, the family and the patient were explained the diagnosis and the prognosis, and the family was also given the information that there is no disease-modifying therapy for this disease. The follow of the patient after six months were poor progressive clinical manifestations and dysphagia, it is difficult to deal like these cases in Somalia, The patient was consulted with a multidisciplinary team for systematic treatment. This is the first case in Somalia to be diagnosed with this illness as far as we are aware, there is no any case report regarding this condition in the literature for Somalia. In low-socioeconomic and low-resource countries, there is a lack of advanced investigations like genetic tests, PET scan (positron emission tomography), and SPECT (single photon emission computed tomography) to diagnose this condition and its differentials. There is no connection between low socioeconomic status and MSA, but diagnosing this condition is challenging in sub-Saharan countries due to a lack of availability for the diagnosis of MSA. Over the course of eighteen years, a retrospective study conducted in North Tunisia revealed that the majority of MSA patients were MSA-P motor-subtype, primarily with PIGD-phenotype, illness duration, and APOEε4 carrying status, defining a more altered cognitive phenotype, also similar case reports as differential diagnosis being performed in somalia [21,22].

Conclusion

Multiple system atrophy is a rare neurodegenerative disease that has rapidly progressive clinical manifestations and is poorly responsive to levodopa. It needs highly advanced imaging and genetics for diagnosis. Our case was a 54-year-

old male who presented with parkinsonism, atakxia, and dysarthria. After a close neurological and MRI brain evaluation, he was diagnosed with multiple system atrophy cerebellar type, which is the first time to have this diagnosis in Somalia, which is a low-resource country.

Patient consent

Written informed consent was obtained from the patient for this case publication and is available to the corresponding author in need.

Ethical consideration

No ethical approval is required for the publication of case reports from our hospital.

Author contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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