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Case Report

Shy-Dragger Syndrome – An underdiagnosed sad reality?

Vasco Pinto Neves[1]*, Maria Pacheco[2], Maria Inês Silva Gonçalve[3], Joao Rodrigues[4], Inês Salvado[5], Jessica Fidalgo[6] and Pedro Daniel Carneiro Ventura[7]

[1]Medical Doctor, Master in Medicine by Faculdade Ciências da Saúde, Universidade da Beira Interior, Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

[2]Medical Doctor, Lincensed in Medicine By Universidad de Salamanca, Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

[3]Medical Doctor, Master in Medicine by Faculdade Ciências da Saúde, Universidade da Beira Interior, Hospital Pêro da Covilhã, Centro Hospital Cova da Beira, Alamêda Pêro da Covilhã, Covilhã, Portugal

[4]Medical Doctor, Master in Medicine By Instituto de Ciencias Biomédicas Abel Salazer, Universidade do Porto. Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

[5]Medical Doctor, Master in Medicine by Faculdade Ciências da Saúde, Universidade da Beira Interior, Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

[6]Medical Doctor, Master by Faculdade de Medicina De Lisboa, Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

[7]Medical Doctor, Master in Medicine by Faculdade de Medicina da Universidade do Porto, Hospital Sousa Martins, Unidade Local de Saúde da Guarda, Av. Rainha D. Amélia, Guarda, Portugal

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***Corresponding author:** Vasco Pinto Neves, Medical Doctor, Internal Medicine, Faculty of Health Sciences, University of Beira Interior, Portugal, Tel: 00351925348055; E-mail: vbenj@icloud.com

ORCID: https://orcid.org/0000-0002-1947-2716

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Abstract

Multiple System Atrophy (MSA) regards a group of neurodegenerative diseases sharing the same physiopathology. It is a rare group of diseases and often represents a diagnostic challenge for clinicians. Mild symptoms are present at the onset of the disease and are often neglected by patients. The case report describes a 62-year-old female with multiple episodes of syncope with 3 months evolutions. Anamnesis revealed sleep apnoea and urinary incontinence. In order to perform complementary exams, the patient was admitted to the medical ward. Autoimunnity results were negative discarding this etiology; 24-hour arterial pressure monitoring revealed a severe fluctuation of tensional values capable of explaining syncope. Other exams revealed no pathological alterations responsible for the patient complaints. Multiple System Atrophy diagnosis was made after exclusion of other possible aetiological causes for the patient's symptoms. More prevalent diseases such as Parkinson's Disease, atypical Parkinsonic syndromes, and pure autonomic failure, among others may replicate the same symptoms. Diagnosing the patient with Multiple System Atrophy represented a challenge because of its rarity and clinical complexity. Being a disease with poor outcomes and representing necessary lifestyle changes to the patients and family life, an early and exact diagnosis may provide time and more life quality, within the disease limitations.

Internists are often presented with complex patients being obligated to gather all pieces, and put them together, so that de puzzle may be deciphered.

Learning points

- Multiple systems atrophy is a rare incapacitating disease and it is believed to be underdiagnosed;

- Complex diseases often present mild nonlimiting symptoms that patients might tend to neglect.

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Case description

Introduction

Multiple System Atrophy (MSA) regards a group of neurodegenerative diseases sharing the same physiopathology. They are characterized by variable degrees of dysautonomia, cerebellar dysfunction, Parkinsonism, and cortico-spinal dysfunction [1,2]. Mental status is often preserved [3]. It is a rare condition, with the onset of symptoms typically between 50 and 55 years [4].

The cause of MSA is unknown [5]. Multiple mechanisms have been postulated even though it remains under investigation.

The diagnosis of MSA is based on clinical features. No laboratory or imaging studies are diagnostic.

Case report

The 62-year-old Caucasian female was referred to Internal Medicine consultation for multiple episodes of syncope. Relevant medical history included sleep apnoea, vertigo, depression, and urinary incontinence. Chronically medicated with fluoxetine 20mg twice daily, trimetazidine 35mg daily, and nocturnal CPAP. Upon asking the patient mentioned no relevant familiar medical history.

In the first consultation patient presented with complaints of paroxysmic episodes of generalized weakness associated with dizziness and blurred vision, sometimes ending with fainting. The patient reported a crisis with 3 months evolution. Clinical examination revealed obesity (Body Mass Index of 31%), slurred voice with a minor degree of dysarthria, intention tremor with severe dysgraphia, positive Romberg signal, normal arterial pressure in decubitus associated with orthostatic hypotension (Mean arterial pressure lower than 50 mmHg) with severe dizziness associated with postural changes.

The medical team decided to admit the patient to the medical ward for further exams, which included the results summarized in the following (Tables 1-3).

Before the results of complementary diagnostic exams and clinical manifestations with clear signs of Parkinsonism, cerebellar dysfunction, and dysautonomia, the medical team responsible for the patient made the diagnosis of Multiple System Atrophy, namely Shy Dragger Syndrome Subtype due to the predominance of dysautonomia. The patient was medicated with an Alfa-adrenergic agonist and Beta-Blockers for orthostatic hypotension, with only partial clinical response. Later, the patient was referred for medical consultation with a neurologist and a physiatrist.

Table 1: Diagnostic tests performed and their results.

Diagnostic test	Result
Hemogram	Normal
Biochemistry	Sedimentation speed 1st hour: 80mm/s; no further alterations
Thyroid function	Normal
Cortisol (morning plus 24 hours)	Normal
Renin-angiotensin-aldosterone axis hormones	Normal
Metanephrines	Normal
HIV; HBV; HCV serologies	Negative
B12 vitamin; Folic acid; plasmatic iron	Normal
Autoimmunity (antineutrophil antibodies; anti-neutrophil cytoplasmic antibodies; Anti-ganglioside antibodies)	Negative

Table 2: Cardio-vascular Exams realized and their results.

Cardio-vascular Exams	Result
ECG	Normal
Echocardiogram	Minor diastolic dysfunction; no other pathological alterations
Carotid doppler	Normal
24-hour arterial pressure analysis	Severe daily fluctuation of tensional values. Max Sistolic pressure of 158mmHg, min systolic pressure of 64mmHg. Max diastolic pressure one 110mmHg, min diastolic pressure of 40mmHg)
24hour Holter monitoring	Sinus rhythm during the hole monitoring. Minimum cardiac frequency of 46bpm, and a maximum frequency of 104bpm. Presence of rare supra ventricular extrasystoles
Tilt Test	No pathological findings

Table 3: Neurological Exams realized and their results.

Neurological exams	Results
Cranioencephalic CT	Minor diffuse pattern of microangiopathic leucoencefalopathy
Cranioencephalic MRI	Scattered hyperintense foci suggest ischemic dots secondary to macroangiopathic lesions. Mild atrophy of structures in the posterior fossa.
Electroencephalogram	No pathological alterations

Discussion

Multiple System Atrophy diagnosis is made after exclusion of other possible aetiological causes for the patient's symptoms. More prevalent diseases such as Parkinson's Disease, atypical Parkinsonic syndromes, and pure autonomic failure, among others may replicate the same symptoms. Differentiation between the different pathologies is a medical challenge, and the definitive diagnosis may be available only after post-mortem biopsies. Diagnosing the patient with Multiple System Atrophy represented a challenge because of its rarity and clinical complexity. Internists are often presented with complex patients being obligated to gather all pieces, and put them together, so that de puzzle may be deciphered. Literature regarding the disease often describes much earlier signs of the developing disease; in this case report, sleep apnoea and urinary incontinence were signs of such.

Treatment options are limited and directed only towards symptoms. Regarding treatments that alter the course of the illness, no medications are available. The patient developed severe symptoms of dysautonomia and died within two years. No autopsy was performed and therefore there is no information regarding brain biopsy.

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References

1. Consensus statement on the definition of orthostatic hypotension, pure autonomic failure, and multiple system atrophy. The Consensus Committee of the American Autonomic Society and the American Academy of Neurology. Neurology. 1996 May;46(5):1470. doi: 10.1212/wnl.46.5.1470. PMID: 8628505.

2. Stefanova N, Bücke P, Duerr S, Wenning GK. Multiple system atrophy: an update. Lancet Neurol. 2009 Dec;8(12):1172-8. doi: 10.1016/S1474-4422(09)70288-1. PMID: 19909915.

3. Schatz IJ. Shy-Drager syndrome. N Engl J Med. 1985 Feb 7;312(6):378-9. doi: 10.1056/NEJM198502073120617. PMID: 3969092.

4. Tison F, Yekhlef F, Chrysostome V, Sourgen C. Prevalence of multiple system atrophy. Lancet. 2000 Feb 5;355(9202):495-6. doi: 10.1016/S0140-6736(00)82050-4. PMID: 10841152.

5. Wenning GK, Stefanova N, Jellinger KA, Poewe W, Schlossmacher MG. Multiple system atrophy: a primary oligodendrogliopathy. Ann Neurol. 2008 Sep;64(3):239-46. doi: 10.1002/ana.21465. PMID: 18825660.

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