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FXTAS: A challenging diagnosis

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Abstract

We present a case of a 56 year old man with a long standing history of anxiety and depression that evolved in an uncommon syndrome that consisted in tremor and overall motor dysfunction being finally diagnosed of Fragile X-associated tremor/ataxia syndrome.

Keywords

FXTAS; neurodegenerative disorder; tremor; anxiety.

Abbreviations

FXTAS: Fragile X-associated tremor/ataxia syndrome.

Introduction/background

Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) is a progressive neurodegenerative disorder that happens among the premutation carriers of the FMR1 (Fragile X mental retardation 1) gene (also associated with Fragile X Syndrome (FXS) of late onset [1].

FXTAS is characterized by the development of both cognitive signs and motor symptoms. Patients usually show the first symptoms in their early sixties, and the rate of progression increases with age. Being the incidence described of 40% of male premutation carriers having developed the syndrome by the age of 70 years old, while 75% after the age of 80 [2].

Due to the nonspecific nature of its symptoms the diagnosis is complicated and requires a high index of clinical suspicion [3].

Case Report

A 56-year-old man, working as a high school teacher, without any known allergies nor any other relevant pathologies, presented with a 6 month history of anxiety and depression, asthenia and anhedonia,

preventing him from reconciling a normal family and work life. Treatment was initiated with Venlafaxine and Lorazepam, initially reporting a slight symptomatic improvement, but after 5 months of treatment the patient started with difficulty articulating words. Not just the dysarthria, but he also presented with difficulty writing with his right hand, as well as instability when walking.

The physical examination showed a normal posture of the patient, bradykinesia of the upper right arm and a positive froment´s sign of the right hand. Mild scanning dysarthria was present. Tendon reflexes were preserved and symmetrical. Tandem walk rightly performed.

A cranial CT scan was performed, where there were no relevant findings and the patient was referred to the neurology department.

The MRI performed showed a mild atrophy of the cerebellar foliae without any other findings. In the FP-CIT SPECT a decreased transporter density was seen in the left putaminal region and the lack of alterations in the Cardiac-MIBG SPECT discarded the diagnosis of Lewy body dementia.

Laboratory test results showed normal red, white blood cell and platelet counts, liver function, renal function, electrolytes, C-reactive protein and sedimentation rate, and thyroid function. Vitamin A, E and B12, folic acid, tumor markers and all autoinmune antibodies being negative. Blood serology of HIV, shyphilis, CMV (citomegalovirus), Epstein Barr, Herpes and mumps were also negative.

A genetic study was also performed with positivity for X-fragile premutation (FMR1 gene) compatible with Fragile X Associated Tremor/Ataxia Syndrome (FXTAS).

Treatment with Amantadine, Levetiracetam and escitalopram was initiated with parcial improvement and follow-up being carried out by rehabilitation and speech therapist.

During the next 5 years the patients evolution was deteriorated with a greater instability when walking and clinical signs of salivation and dysphagia with liquids.

In the end, the clinical evolution was torpid, from the general motor disability being almost complete and the multiple hospital admissions due to aspiration pneumonia, the patient finally passed away.

Discussion

FXTAS is a neurodegenerative disorder, progressive and of late onset that happens among the premutation carriers of the FMR1 (Fragile X mental retardation 1) gene (also associated with Fragile X Syndrome (FXS) [1].

The clinical features that should be taken into account as to suspect the syndrome are in particular men around the age of 50 presenting tremor, ataxia and parkinsonian traits, or multiple of these symptoms together. Women, on the other hand, have milder symptoms, being the number of cases described in the literature in women far less than in men [3].

Due to its similarity with other neurological disorders FXTAS is often underdiagnosed [4].

In order to diagnose the syndrome its important to perform a detailed family history and a genetic testing for FMR1 repeat expansion when is highly suspected. There is presently no targeted treatment

available for FXTAS, but clinical trials are being carried out. Identified carriers should modify their lifestyle in order to either prevent harmful environmental exposure and somehow delay FXTAS symptoms [5].

Declarations

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Declaration of competing interest: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Confidentiality of the data: The authors declare that they have followed the protocols of their work center on the publication of patient data.

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