RESEARCH WORK

Spastic Paraplegia Type 7 in the Differential Diagnosis of Cerebellar Ataxia and Late-onset Cervical Dystonia. Case Report

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ABSTRACT

Autosomal recessive spastic paraplegia is a heterogeneous group of heredodegenerative diseases that present spasticity, muscle weakness, progressive cerebellar ataxia and less frequently involuntary movements. The present case refers to a 44-year-old male patient presenting a clinical picture of insidious and progressive onset 10 months before the evaluation and characterized by spasticity, muscle weakness, cerebellar ataxia and cervical dystonia. The brain magnetic resonance imaging study showed marked cerebellar atrophy. Laboratory tests and cerebrospinal fluid results were within normal limits. The genetic study showed a homozygous mutation in the SPG7 gene. Symptomatic treatment was instituted with partial improvement of symptoms.

Key words: Spastic paraplegia type 7, cerebellar ataxia, cervical dystonia, case report.

INTRODUCTION

Late-onset ataxias represent a large and heterogeneous group of diseases. Patients rarely have defining clinical features, and many remain classified as idiopathic despite extensive clinical, metabolic, and genetic investigations. Hereditary spastic paraplegia type 7 (SPG7) is a cause of adult-onset ataxia, autosomal recessive, heredodegenerative, with spasticity, muscle weakness, progressive cerebellar ataxia, and less frequently involuntary movements.

METHODS

The report of this case was submitted and approved by the ethics committee of Universidade Metropolitana de Santos.

CASE PRESENTATION

The present case refers to a 44-year-old male patient who presented a clinical picture of insidious and progressive onset 10 months after the evaluation and which was characterized by lack of balance, muscle weakness, and involuntary posture in the neck. The neurological examination showed spastic tetraparesis (muscle weakness strength grade 4), signs of pyramidal release in lower members, axial and appendicular ataxia, nystagmus evoked by the gaze position, dysdiadochokinesia, dysarthria and cervical dystonia.

The presence of intellectual deficit was also observed and with intellectual impact. Several clinical and complementary neurological evaluations were performed, including graphic

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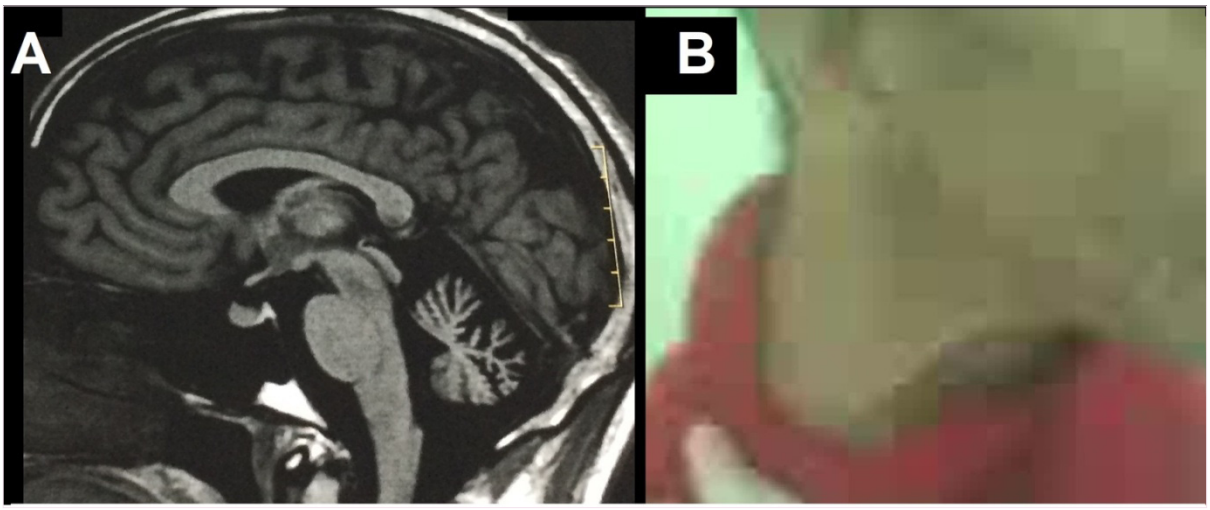

Figure: A- FLAIR/Sagittal Brain MRI- Global cerebellar atrophy. B- Cervical dystonia

(electroencephalogram) and imaging (brain and spinal cord magnetic resonance imaging) with marked cerebellar atrophy. (Figure)

Laboratory tests and cerebrospinal fluid results were within normal limits.

The genetic panel for ataxias study was performed and showed a homozygous pathogenic variant Chr16:89.595.896 CTG>C in the SPG7 gene. This variant promotes the replacement of the aminoacid valine at position 258 by glycine and changes the reading matrix from this point onwards, with the consequent creation of a premature stop codon (p.Val258Gly fs*30).

This pathogenic variant is present in patients with hereditary spastic paraplegia, because it encodes the paraplegin protein as well as promotes early interruption of protein translation. Autosomal recessive spastic paraplegia 7 pathogenic variants can manifest as pure or complicated spastic paraplegia. Clinically, it is characterized by the onset of walking difficulties in general in the second or third decades of life, with weakness and spasticity, frequent loss of vibratory sensitivity, urinary incontinence and the presence of additional cerebellar signs such as dysphagia, dysarthria, ataxic gait and nystagmus. It is a condition with an autosomal recessive pattern of inheritance; however cases with an apparent autosomal dominant pattern, associated with some specific mutations, are also reported. (1-3)

The finding of the pathogenic variant in the SPG7 gene was postulated to be associated with this patient's clinical condition.

Multi professional treatment was performed with physiotherapy, speech therapy, occupational therapy and psychology. Drug treatment was instituted with baclofen at a dose of 20 mg daily and clonazepam 2.0 mg daily with an adequate response.

Clinical and rehabilitation treatment resulted in partial improvement of neurological symptoms.

CONCLUSION

Spastic paraplegia type 7 is a challenge due to the heterogeneity of clinical presentation. With the advent and development of neurogenetics, many etiologies were then better understood. The study of the SPG7 gene should be encouraged in these patients, especially in individuals with spasticity, muscle weakness, progressive cerebellar ataxia and involuntary movements.

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