REVISTA NEUROCIÊNCIAS

Relato de Caso

Oromandibular chorea as the initial manifestation of Juvenile Huntington's Disease

Coreia oromandibular como manifestação inicial de Doença de Huntington juvenil

Corea Oromandibular como manifestación inicial de la enfermedad de Huntington Juvenil

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Resumo

Objetivo. Apresentação da Doença de Huntington Juvenil com coreia oromandibular como manifestação inicial. **Método.** Relato de caso e breve revisão da literatura. **Resultados**. Apesar do fenótipo variante clássico de Westphal sem distúrbios do movimento hipercinético na doença de Huntington juvenil, alguns pacientes podem apresentar coreia como apresentação inicial da JHD. **Conclusões.** A fenomenologia hipercinética não descarta JHD no contexto de achados sugestivos de neuroimagem e história familiar de HD, apesar da clássica apresentação rígida-acinética da variante Westphal.
Unitermos. Doença de Huntington Juvenil

Abstract

Objective. Description of Juvenile Huntington's Disease with oromandibular chorea as the initial manifestation. **Method.** Case report and brief literature review. **Results.** Despite classical Westphal's variant phenotype without hyperkinetic movement disorders in Juvenile Huntington's Disease, some patients could present with chorea as initial presentation of JHD. **Conclusions.** Hyperkinetic phenomenology does not rule out JHD in the context of suggestive neuroimaging findings and familiar history of HD despite classic akinetic-rigid presentation of Westphal's variant.
Keywords. Juvenile Huntington Disease

Resumen

Objetivo. Descripción de la Enfermedad de Huntington Juvenil con corea oromandibular como manifestación inicial. **Métodos.** Presentación de un caso y breve revisión de la literatura. Resultados. A pesar del fenotipo variante de Westphal clásico sin trastornos del movimiento hipercinético en la enfermedad de Huntington juvenil, algunos pacientes pueden presentar corea como presentación inicial de JHD. **Conclusiones.** La fenomenología hipercinética no descarta JHD en el contexto de hallazgos de neuroimagen sugestivos y antecedentes familiares de HD a pesar de la clásica presentación acinética-rígida de la variante de Westphal**. Palabras clave.** Enfermedad Juvenil de Huntington

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INTRODUCTION

Huntington's disease (HD) is an inherited progressive neurodegenerative disorder caused by a cytosine-adenine-guanine (CAG) trinucleotide repeat expansion in the *HTT* gene. Patients with Juvenile Huntington's disease (JHD), when disease presents before 20-years-old, usually have minimal or no chorea, the classical presentation of Westphal's variant[1]; juvenile-onset patients are more prone to parkinsonism, myoclonus, and behavioral problems[2,3].

We present a case of a children with prominent features of choreic movements at onset, an unusual presentation of JHD.

CASE REPORT

We report a case of a 10-year-old boy who was referred due oromandibular chorea since 5-years-old. After 2 years, he had dysarthria and poor school performance, combined with agitation and irritability. Ethics Comittee approved this study under protocol number 09144819.1.0000.5292.

The patient was born in Brazil, from non-consanguineous parents, and had normal early developmental milestones and cognitive function. His

previous medical history was unremarkable. His father has genetically confirmed Huntington's disease (47 CAG repeats) at age of 41-year-old.

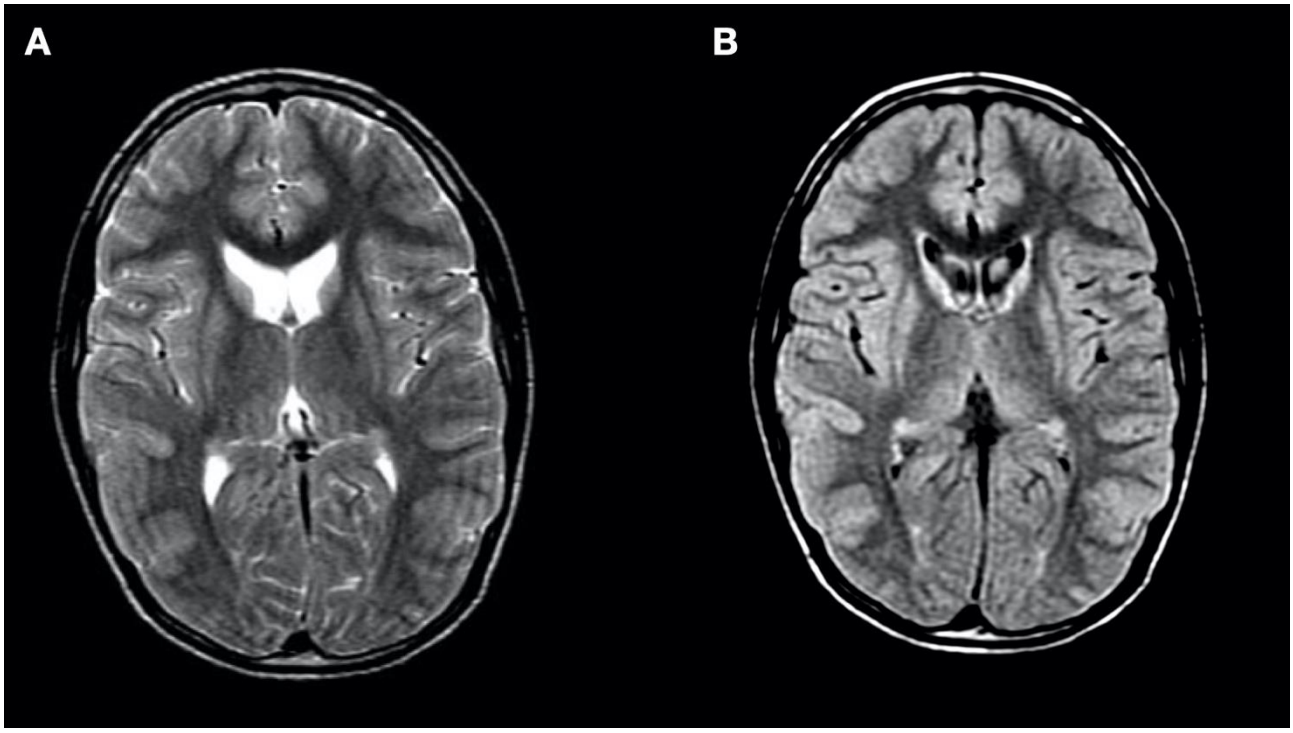
His neurologic examination showed mild dysarthria; assessment of cranial nerves was unremarkable. Motor examination was marked by uncontrolled motor impersistence and choreic movements comprising predominantly facial muscles and distal extremities (Figure 1). His reflexes were normal and he has not revealed rigidity and bradykinesia. Sensory examination was normal.

Brain magnetic resonance imaging (MRI) showed bilaterally caudate and putamen atrophy (Figure 2). Genetic testing of *HTT* gene disclosed an abnormally expanded 71 CAG repeats allele compatible with juvenile HD.

Figure 1. Neurological examination findings.

A-B, Choreic movements of distal superior extremities; C-D, Oromandibular choreic movements

Figure 2. Juvenile Huntington's disease Brain MRI findings.



A, T2WI Brain MRI and B, FLAIR/T2WI revealing caudate and putamen bilateral and caudate atrophy. Atrophy in JHD is usually more severe than in Adult Huntington's Disease.

DISCUSSION

Juvenile Huntington's disease (JHD), defined as HD with an onset ≤20 years, accounts from 4.81 to 9.95% of all cases of HD and only approximately 20% of JHD have childhood-onset (<10-year-old) as presented in this case. Individuals with a CAG repeat length of >60 usually have JHD, and transmitting parent is frequently the father (~70–80% of cases)[4]. Although there are many similarities with the adult form of the disease, JHD has a clinically distinct presentation, as the pattern tends to be that the bradykinesia, dystonia, and parkinsonian features are prominent at an early stage, while chorea, if present, is less

prominent[1,3]. On this case, nonetheless, the initial, longstanding, and predominant feature was chorea, initially oromandibular, and later progressing to the distal extremities. With language and psychiatric involvements presenting only later on the course of the disease.

This patient was managed with risperidone, and showed great response to it, having had control of the choreic movements.

CONCLUSION

Hyperkinetic phenomenology does not rule out JHD in the context of suggestive neuroimaging findings and familiar history of HD despite classic akinetic-rigid presentation of Westphal's variant.

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