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Case Report

Juvenile-onset Huntington's Disease: A Rare Case Report

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Abstract

Juvenile-onset Huntington's disease (JHD) is a rare autosomal dominant neurodegenerative disorder of the central nervous system characterized by the presence of abnormal involuntary movements, rigidity, and ataxic gait. We are presenting a rare case of a 9-year-old male who was referred to the Radiology Department of Gandhi Medical College and Hamidia Hospital for magnetic resonance imaging (MRI) brain with complaints of progressive impairment of gait, bradykinesia, and marked postural instability for the past 2 years. The patient also had a history of episodes of seizures for 4 years. MRI findings revealed: Atrophy of bilateral caudate nuclei and putamina of basal ganglia.

Key words: Atrophy caudate nucleus, Juvenile-onset Huntington's disease, Putamina

INTRODUCTION

Juvenile-onset Huntington's disease (JHD) is a rare autosomal dominant neurodegenerative disorder of the central nervous system characterized by the presence of abnormal involuntary movements, rigidity, and ataxic gait.

In 1872, George Huntington (1850–1916), a medical practitioner of Pomeroy, Ohio, USA, made the first complete description of this disorder among the population of Long Island in New York State.[1]

The molecular origin of the disease is the cytosine adenine guanine trinucleotide increase in the Huntington gene which is located in 4p16.3 chromosome.[2]

We are presenting a case of a 9-year-old male who was referred to the Radiology Department of Gandhi Medical College and Hamidia Hospital for magnetic resonance imaging (MRI) brain with complaints of progressive impairment of gait, bradykinesia, and marked postural instability for the past 2 years. The patient also had a history of episodes of seizures for 4 years.

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MRI findings revealed:

- Marked atrophy of bilateral caudate head resulting in enlargement of bilateral frontal horns [Figure 1].[3]

It can be quantified by three measurements that were obtained on axial images at the level of the third ventricle: (a) The FH width, representing the distance between the most lateral aspects of the frontal horns; (b) the intercaudate distance (CC), the distance between the most medial aspect of the caudate nuclei; and (c) the IT width, the distance between the inner tables of the calvarium at the level of the CC measurement [Figure 2].

- Frontal horn width to intercaudate distance ratio (FH/CC), which in our case is −0.85 (normal range is 2.2–2.6)
- Intercaudate distance to inner table width ratio (CC/IT), which in our case is 0.26 (normal range is 0.09–0.12); (as the caudate heads are reduced in size, the CC distance will increase and so will the CC/IT ratio).

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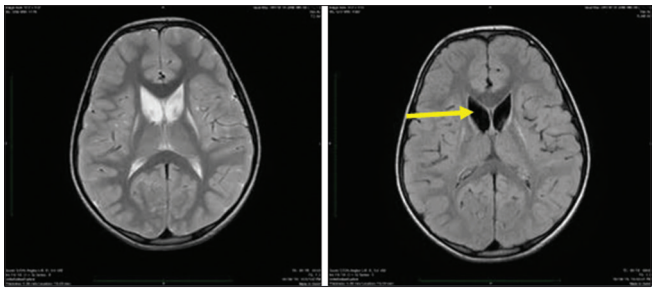

Figure 1: Axial T2 and FLAIR images show bilateral caudate head atrophy [arrow] resulting in enlargement of frontal horns. Frontal horn width to intercaudate distance ratio (FH/CC) is –0.85 [normal range is 2.2–2.6]. Intercaudate distance to inner table width ratio (CC/IT) is 0.26 [normal range is 0.09–0.12], (as the caudate heads are reduced in size, the CC distance will increase and so will the CC/IT ratio)

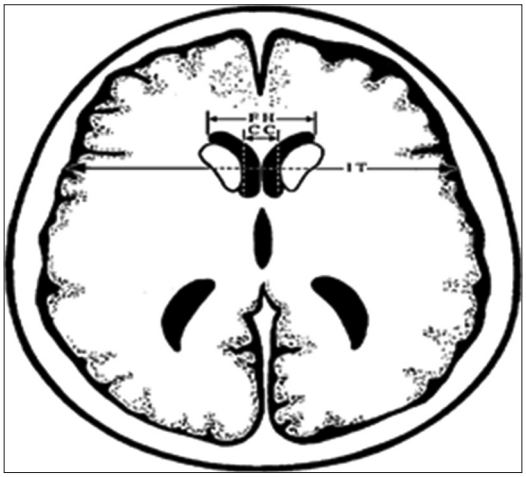

Figure 2: Schematic diagram of an axial view at the level of the third ventricle shows heads of the caudate nuclei at which the intercaudate distance (CC), the frontal horn width, and calvarial inner table width can be measured

- Atrophy of putamina of bilateral basal ganglia with T2/FLAIR hyperintense signals within [Figure 3].
- Rest of the cerebral, cerebellar brain parenchyma, and brainstem was normal [Figure 4], thus ruling out its differentials which are hypomyelination with atrophy of basal ganglia and cerebellum (H-ABC), Leigh and Wilson disease.

Points to Ponder

1. Huntington disease usually occurs in adults (third or fourth decades) and is rare in children; then, it is classified as JHD. Its prevalence in world ranges from 5 to 10/100,000.[4]
2. JHD has a more rapidly progressive course, with death occurring in 7–8 years of disease onset.[4]

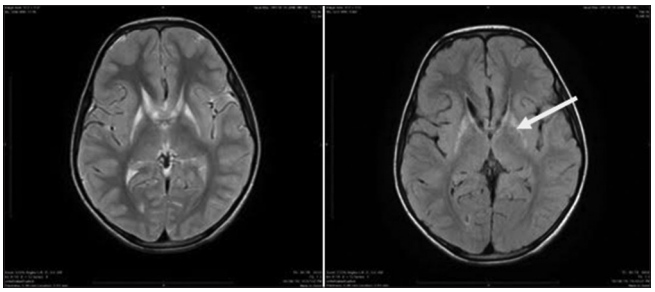

Figure 3: Axial T2 and FLAIR images show bilateral symmetrical hyperintensities in atrophied bilateral putamina [arrow]

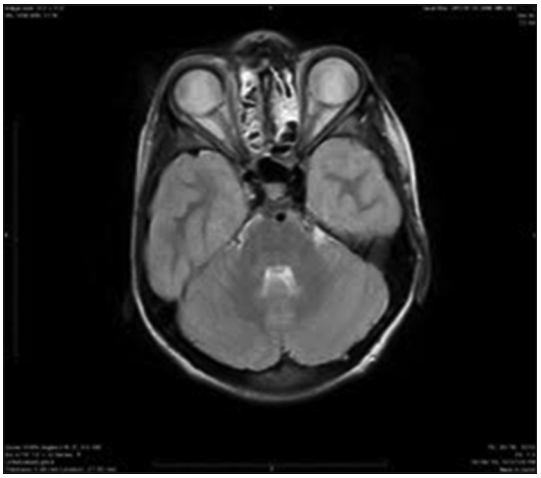

Figure 4: Axial T2 image shows normal cerebellar hemispheres, thus ruling out hypomyelination with atrophy of basal ganglia and cerebellum (H-ABC)

REFERENCES

1. Huntington G. On chorea. Med Surg Report Phila 1872;26:317-21.
2. Gusella JF, Wexler NS, Conneally PM, Naylor SL, Anderson MA, Tanzi RE, *et al.* A polymorphic DNA marker genetically linked to huntington's disease. Nature 1983;306:234-8.
3. Aylward EH, Schwartz J, Machlin S, Pearlson G. Bicaudate ratio as a measure of caudate volume on MR images. AJNR Am J Neuroradiol 1991;12:1217-22.
4. Gonzalez-Alegre P, Afifi AK. Clinical characteristics of childhood-onset (juvenile) huntington disease: Report of 12 patients and review of the literature. J Child Neurol 2006;21:223-9.

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