P67

Atypical sonographic findings in a young man with parkinsonism in Machado-Joseph disease

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Introduction: Machado-Joseph disease (MJD), also known as spinocerebellar ataxia type 3 (SCA3), is an autosomal dominantly inherited cerebellar ataxia caused by CAG trinucleotide repeat expansions in the coding regions of ATXN3 gene [1]. Few studies have performed transcranial sonography (TSC) in SCA 3 patients and found hyperechogenicity of Substantia Nigra (SN) associated with a larger width of third ventricle and lenticular nucleus hyperechogenicity compared to healthy controls [2-3]. The disease is characterized by phenotype variability and several subtypes have been defined [4] that could be related to the atypical echofeatures found in our young patient.

Objective: Case report.

Methods: We studied the case of a 27 years old African man with a 3-years history of gradually progressive slowness of movements and balance difficulty, with referred negative family history for neurological diseases. He underwent full neurological examination and genetic analysis on peripheral blood sample using PCR and capillary electrophoresis.

Results: Neurological examination revealed marked bradykinesia (mainly on the left) and gait ataxia, moderate gaze-evoked horizontal nystagmus and generalized hyperreflexia, without tremor, hypertonia or rigidity. Poor response to L-dopa therapy.
Brain MRI showed cerebellar vermian atrophy with ventricular enlargement.
123I-FP-CIT SPECT revealed a moderate bilateral reduction of dopamine presynaptic transporter levels.
TCS showed third ventricular enlargement and right lenticular nucleus hyperechogenicity. There was no evidence of SN hyperechogenicity.
Genetic analysis demonstrated pathogenetic CAG repeat expansion (67 repeats) in the coding region of ATXN3 gene.

Conclusions: In agreement with previous studies, MJD should be considered in the differential diagnosis of PD-like symptoms. The enlargement of third ventricle but the absence of SN hyperechogenicity found in our patient may suggest the presence of different pathophysiologic substrates correlated to a possible distinctive phenotype, that deserves to be investigated. Furthermore, given the fact that lenticular nucleus hyperechogenicity is associated with dystonia [5], follow-up is indicated regarding a possible development of this symptom.

References:

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