A YOUNG WOMAN WITH THE DIAGNOSIS OF WESTPHAL-VARIANT OF HUNTINGTON'S DISEASE

INTRODUCTION: HD is a genetic disease with an AD transmission due to an expansion of the CAG trinucleotide repeat in the huntingtin gene. Mean age of onset is 40 years, but it varies according to the trinucleotide repeat expansion with an inverse relation between the number of CAG repeats and age of onset (genetic anticipation). CASE REPORT: A 21yo woman with previous normal neurodevelopmental milestones, at the age of 10 started progressive social withdrawal and difficulties in school activities. Thereby, she failed in completing high school education and developed a severe depression with suicide attempt. 9 years later, she noticed progressive bradykinesia. Neurological examination showed myoclonic jerks, facial and distal upper-limb choreiform movements, parkinsonism and oculomotor apraxia. She scored 19/30 on MoCA with no impairment of ADLs. About family history, her father, at 40yo, developed social phobia and obsessive-compulsive symptoms. 3 years later, he developed hand writing issues due to involuntary movements. PROPAEDEUTICS: Basic laboratorial biochemistry, ceruloplasmin, copper and α-fetoprotein levels were within normal ranges. Brain MRI showed prominent bilateral atrophy of the head of the caudate. RESULTS: Genetic testing for HD showed 47 CAG trinucleotide repeats in the huntingtin gene in her father and 61 CAG repeats in the patient. CONCLUSION: Less than 10% of HD cases occur before 20 years of age. Personality and behavioural issues, as well as executive cognitive decline, are usual characteristic symptoms at the disease onset. These patients are usually first evaluated by psychiatrics and the movement disorder appear later in adolescence, often leading to a diagnostic delay.

KEYWORDS: Huntington. Juvenile. Westphal. Chorea. Neuropsychiatric.