9. CLINICAL PARTICULARITIES OF INTELLECTUAL DISABILITY IN BOURNEVILLE-PRINGLE'S DISEASE

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Introduction. Tuberous sclerosis or Bourneville-Pringle's disease is a genetic disorder with an autosomal dominant transmission that variably affects the brain, skin, kidneys, heart, and other organs, characterized by multisystemic lesions. The disease results from mutations in one of the two genes: TSC1 and TSC2, encoding protein hamartin gene and protein tuberin gene. In 67% of patients, the disease resulted from de novo genetic mutations. We report in this article a case of tuberous sclerosis in a 10-year-old female. Physical examination revealed epileptic seizures and intellectual disability, as well as cutaneous manifestations such as fibromatous nodule, hypopigmented macules, and shagreen patch.

Case presentation. A 10-year-old female presented in the emergency department with generalized seizures in association with antiepileptic medication intake, gait disorders, memory, and behavior disorders. She is the second child in the family, born from a physiological pregnancy completed by a eutocic delivery, at term, weighing 3300 g and APGAR score 8/8, without signs of perinatal suffering. Psychomotor development appropriate up to 4 years old. It was identified that the first child in the family, a girl, has similar manifestations, but in a lighter form. The younger brother is healthy. The first symptoms identified at the age of 4 were seizures. The Neurologist establishes the diagnosis of epilepsy and generalized seizures initiating subsequently antiepileptic therapy with Valproic acid at age-appropriate doses. Consulted by a Medical Geneticist, the diagnosis of tuberous sclerosis was suspected based on clinical criteria: intellectual disability, generalized seizures, numerous fibromatous nodules in the thoraco-lumbar region.

Discussion. Tuberous sclerosis is characterized by the classic triad: cutaneous manifestations, epilepsy and intellectual disability. Clinical manifestations can range from fruste forms with minimal symptoms to severe forms with multi-organ damage. Epileptic seizures are the most common extracutaneous symptom of tuberous sclerosis. Their incidence is 80-90% often with onset in the first year of life, represented by infantile spasms or various types of partial or generalized seizures. The onset of epilepsy usually occurs between the ages of 4-7. The evolution is, in 25-50% of cases, towards treatment-resistant epilepsy. Intellectual disability is variable, from moderate to severe, associated with multiple behavioral problems, such as: hyperactivity, attention deficit disorder, aggression, sleep disorders, autism spectrum disorders. The treatment of neurological manifestations is mainly focusing on managing generalized seizures.

Conclusion. Tuberous sclerosis is a multisystemic disease, characterized by a variety of signs and symptoms that make it difficult for establishing an early diagnosis, in some cases, the diagnosis is delayed for long periods of time. The detailed consultation of the patient by the medical team ensures the establishment of a definite diagnosis of tuberous sclerosis. Early diagnosis is very important for complete clinical and radiological evaluation, continuous monitoring of clinical manifestations, family planning, genetic counseling, as well as reducing the rate of morbidity and mortality.