P-162 - DISORDER OF TETRAHYDROBIOPTERIN CAUSED BY DEFICIENCY OF DIHIDROPTERIDINE REDUTASE (DHPR)

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INTRODUCTION: Hyperphenylalaninemas are manifestations of metabolic errors usually related to amino acid metabolism. Five defects that can lead to hyperphenylalaninemia are known: one in the enzyme phenylalanine hydroxylase, causing phenylketonuria, and in four enzymes involved in the synthesis or regeneration of tetrahydrobiopterin (BH4), an enzymatic cofactor involved in the synthesis of dopamine, serotonin, tyrosine, nitric oxide and glycerol. BH4 deficiencies account for approximately 2% of cases of hyperphenylalaninemia with a worldwide prevalence of 1/1,000,000 live births and are designated according to underlying enzyme deficiency and may lead not only to hyperphenylalaninemia states but also to neurological symptoms and signs of neurotransmitter deficiency. These disorders promote a progressive deterioration of the neurological function, and cannot be avoided by phenylalanine restrictive diet alone.

OBJECTIVE: Case report of a patient with DHPR deficiency, presenting difficult diagnosis and therapeutic limitation with severe clinical and laboratory neurological alterations.

MATERIALS AND METHODS: This information was obtained by means of a medical record review, interview with the patient's parents, physical examination of the child, photographic record of neuroimaging, and literary review using Medline / Pubmed and specialized literature.

RESULTS: JPS, 7 years, consanguineous parents. Prenatal and delivery without complications. Normal exams. Onset symptoms at 2 months of age - seizures, start of anticonvulsant. Normal MRI. 1 year: significant delay in psychomotor development. Change RM Definitive diagnosis: 1 year and 5 months. Brain CT showing absence of cerebral calcifications. Specific treatment inception: 1 year and 5 months: L dopa / carbidopa and low doses of folinic acid due to adverse reactions, reaching ideal doses and association with 5 hydroxytryptophan with 1 year and 7 months. Sapropterin dihydrochloride (KUVAN) - important adverse reactions, therapeutic limitation. 6 years old MRI with typical signs of deposit leukoencephalopathy and severe DNPM changes even with specific treatment.

CONCLUSIONS: The reported case and other publications reveal the rarity of studies related to this pathology, the scarcity of methods of early diagnosis, besides evidencing the late institution treatment consequences and the current therapy limitation, with severe damage to the patient’s psychomotor development.