P-110 - AADC DEFICIENCY IN BRAZIL: CLINICAL AND BIOCHEMICAL FINDINGS OF A RARE NEUROMETABOLIC DISORDER

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BACKGROUND: Disorders of monoamine neurotransmitter metabolism have been increasingly recognized. These compounds have numerous roles including modulation of psychomotor function; hormone secretion; cardiovascular, respiratory, and gastrointestinal control; sleep mechanisms; body temperature; and pain. Aromatic L-amino acid decarboxylase (AADC) is central in the synthesis of biogenic monoamine neurotransmitters. AADC deficiency is a severe neurometabolic disorder, usually underrecognized, presenting early in life with hypotonia, hypokinesia, oculogyric crises, autonomic dysfunction, dysphoric mood, and sleep disturbance. OBJECTIVES: To describe a cohort of 5 Brazilian patients with AADC deficiency. MATERIALS AND METHODS: Clinical, biochemical and radiological data retrospective analysis were reviewed, including the following items: demographic characteristics, genetic profile, and clinical manifestations. RESULTS: All patients showed normal body weights in the first few months of life. Their growth, however, began to slow down at the end of the first year, and their weight gain was minimal between 1 and 4 years of age, although all of them were treated with a combination of pyridoxine, dopamine agonists, and monoamine oxidase inhibitors. Neurological symptoms became evident in all patients during the first 6 months of life. Intellectual disability, truncal hypotonia, hypokinesia and hypomimia, dystonic movements and typical oculogyric crises in all patients. None of our patients had full head control, defined by the ability to hold their head upright in the sitting position, at their termination point in the study. CONCLUSIONS: AADC deficiency is a neurotransmitter disorder with features presenting as early as in the first month of life. Hypotonia, hypokinesia, oculogyric crises and autonomic are clinical hallmarks of the disease and may lead to clinical suspicion. Patients may show a number of movement disorders, most frequently dystonia. Diurnal fluctuation and improvement of symptoms after sleep is characteristic. The severity of the clinical phenotype is variable, but the majority of patients show minimal motor development in the absence of treatment.