

Theses

Contribution to clinical characterization of autosomal recessive hereditary ataxias (Abstract)*. Theses. São Paulo, 2009.

Emília Katiane Embiruçu**

Autosomal recessive hereditary ataxias belong to a group of heterogeneous disorders, for which detailed clinical evaluation, ancillary exams, and sometimes, genetic tests, are required for diagnosis. After literature review, an algorithm was built to help the investigation of this group. The objective of this thesis is to present the results of investigation of three forms of recessive ataxias: [1] Joubert syndrome is a condition characterized by early hypotonia, developmental delay, ataxia and neonatal respiratory disturbances or abnormal eye movement. It has a wide clinical spectrum and is a genetically heterogeneous. Renal, hepatic and retina abnormalities are often seen. A combination of midline cerebellar vermis hypoplasia, deepened interpeduncular fossa, and thick, elongated superior cerebellar peduncles gives to the axial view of the midbrain an appearance of a molar tooth at brain magnetic resonance image (MRI) study. Molar tooth sign is considered as obligatory radiologic criteria to diagnosis. In this study we present a series of five patients that have clinical and radiologic criteria to Joubert syndrome and a large phenotypic variability: two children have a pure form (subgroup 1), one child has an associated retinopathy (subgroup 2), the other has Leber congenital amaurosis and kidney abnormalities (subgroup 4), and another has chorioretinal coloboma and hepatic abnormalities (subgroup 5); [2] Ataxia with vitamin E deficiency, which has a phenotype similar to Friedreich ataxia but slowest progression, is characterized by low levels of serum α -tocopherol and is treatable with vitamin E. This ataxia is common in South Italy and North Africa, but was not reported in Brazil. Four patients from two different families were studied. Three of them have typical clinical features and hands dystonia, a probably underreported feature which might help its distinction from Friedreich ataxia. The other case was identified in a pre-symptomatic stage, after family investigation. After five years of treatment with vitamin E, subtle balance disturbance was still present. The remaining three patients improved with vitamin E supplementation and disease progression

stopped; [3] Cerebrotendinous xanthomatosis (CTX) is a disorder of cholesterol metabolism, characterized by reduction of bile acid synthesis and accumulation of cholestanol, a toxic metabolic. Congenital or juvenile cataract and chronic diarrhea are early manifestations. Cerebellar ataxia, spastic paraplegia, cognitive impairment and tendinous xanthomatosis are also seen. Brain MRI T2-weighted and FLAIR sequences disclosed dentate nucleus hypersignal, a quite feature in CTX. Three patients from two different families, with clinical and radiologic features were studied. In all, serum cholestanol was elevated. MRI spectroscopy demonstrated in cerebellum a peak in 1.2-1.4 ppm, which is a possibly a lipid, not previously described. Treatment with chenodeoxycholic acid improved their gait.

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** Address: Rua Guilhermino de Freitas Jatobá 103/1104 - 40296-320 Salvador BA - Brasil. (E-mail: ekeleao@yahoo.com.br)

Epidemiologic study about restless legs syndrome and migraine (Abstract)*. Dissertation. Ribeirão Preto, 2010.

Karen dos Santos Ferreira**

Restless legs syndrome (RLS) is a sensorimotor disorder with prevalence between 2.5% and 10% in white population. The pathophysiology of RLS involves a dysfunction of the dopaminergic neurotransmitter system. Migraine attacks may also be influenced by dopamine. We developed a study to evaluate a possible association between migraine and RLS. Patients were recruited among the employees of the Clinic Hospital - Medical School of Ribeirão Preto - University of São Paulo, Brazil. We interviewed employees with migraine (n=72) and a control group without migraine (n=72) matched by sex and age, aiming to diagnose RLS according to the International RLS Study Group criteria. We registered clinical and demographic data in a structured form, and all subjects answered the Beck's Depression Inventory. RLS frequency was significantly higher in patients with migraine than in control subjects [25% vs 8%; p=0.01; odds ratio 3.67 (1.36;