

## Theses

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

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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  $\alpha$ -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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### **Epidemiologic study about restless legs syndrome and migraine (Abstract)\*. Dissertation. Ribeirão Preto, 2010.**

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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;

9.88)]. There was no significant association between migraine, RLS and other comorbidities like diabetes, obesity, anemia and drugs used in that study. The type of migraine (with aura or not) and family history did not differ between RLS and control groups. Depression score as measured by Beck's Inventory was more frequent in migraine patients with RLS ( $p=0.04$ ).

**Key words:** migraine, restless legs syndrome, dopamine.

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## Preclinical evidence of antinociceptive action of 3-phenyl-5-(4-ethylfenil)-imidazolidine-2,4-dione in psychopharmacological studies (Abstract)\*. Dissertation. João Pessoa, 2011.

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Imidazolidine derivatives are synthetic products with many different therapeutic applications. The 3-phenyl-5-(4-ethylphenyl)-imidazolidine-2,4-dione (IM-3), recently synthesized from amino acid was selected for psychopharmacological studies. The study has began with screening and behavioral pharmacology of the LD<sub>50</sub> determination. In the screening results indicate a depressant activity on CNS and from the LD<sub>50</sub> doses were chosen for subsequent tests with 50, 100 and 200 mg/kg intraperitoneally. In the next step, methodologies to evaluate the specific antinociceptive activity were used. The first was the writhing induced by acetic acid; afterwards, the formalin test and finally the hot plate test, which is specific for the central antinociceptive activity. In the three methodologies used, the IM-3 showed to be effective in the writhing test by acetic acid at a dose of 200 mg/kg which increased both the latency to the onset of writhing and reduced the number of writhing in the control group and in the formalin test at doses of 100 and 200 mg/kg decreased the time of the paw lick in the second phase of testing. Therefore, from these experimental data, it is possible to infer that the IM-3 has antinociceptive activity of the anti-inflammatory type.

**Key words:** imidazolidine derivatives, psychopharmacology, antinociceptive, anti inflammatory activity.

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\*\*Evidências pré-clínicas da ação antinociceptiva do 3-fenil-5-(4-etilfenil)-imidazolidina-2,4-dione em estudos psicofarmacológicos (Resumo). Dissertação de Mestrado. Centro de Ciências da Saúde da Universidade Federal da Paraíba, João Pessoa PB (Área: Farmacologia). Orientadora: Liana Clébia Soares Lima de Moraes, Co-Orientador: Reinaldo Nóbrega de Almeida. \*Address: Av. Umbuzeiro 1237/402 - 58038-182 João Pessoa PB - Brasil. (E-mail: qronaldo@ig.com.br)

## Hemispheric assymetry of abnormal focal EEG findings (Abstract)\*. Theses. Recife, 2011.

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**Background:** Left and right cerebral hemispheres are morphologically similar. Focal EEG abnormalities should appear with an equal frequency in both of them.

**Objective:** To find out if there is an asymmetry for focal EEG abnormalities in a retrospective study of a series of EEGs.

**Method:** We retrospectively studied 10,408 EEGs from April 2001 to April 2010, separated by age and gender to estimate the frequency of left-sided versus right-sided focal abnormalities. Associated clinical features were also accessed.

**Results:** Discharges were more prevalent in left cerebral hemisphere, in temporal lobe. A stronger lateralization was found among adults. Right-sided discharges occurred more in frontal lobe. Slow waves were more prevalent in left cerebral hemisphere and among adults. Among left-sided slow waves group, women were more prevalent and men, among right-sided group. Left-sided slow waves were more prevalent in temporal and parietal lobes and right-sided, in frontal and occipital lobes. Epilepsy occurred more in patients with focal discharges. Right-sided slow waves were more related to epilepsy and left-sided, to headache.

**Conclusion:** There were significant differences between cerebral hemispheres on focal EEG abnormalities, suggesting that cerebral asymmetry which must result from different specificities.

**Key words:** lateralization, EEG, sharp waves, slow waves.

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