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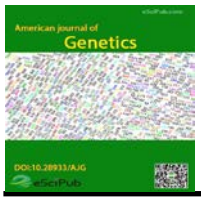

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Genetic Analysis of the Clinical Manifestations of Lipofuscinosis (Batten Syndrome)

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ABSTRACT

Introduction: Neuronal Ceroid Lipofuscinosis (NCL) is a group of neurodegenerative genetic diseases characterized by the accumulation of lipid pigment in neuronal lysosomes and other tissues. Batten syndrome (BS) is the juvenile form of this group, beginning in childhood, with its primary symptomatology manifested between 5-10 years of age, with progressive visual loss and decreased intellectual capacity. Objectives: To correlate genotype and phenotype of Batten Syndrome. Methodology: Articles published between 2000 and 2010 were pre-selected through the PubMed and SciELO databases, using the descriptor: neuronal ceroid lipofuscinoses. Twenty texts were initially analyzed, of which ten were selected after the study of titles and abstracts. Considering inclusion and exclusion criteria, only six were included in the review because they referred to the BS theme in the descriptors and abstracts. Results and Discussion: BS, an inherited autosomal recessive disease, results from the CLN3 gene mutation located on the short arm of chromosome 16p12.1, interfering with the function of the battenin protein. Clinical manifestations begin with dementia, changes in visual acuity, as well as speech disturbances, slow decline in cognitive functions and epilepsy. Motor alterations are more common in the adolescence, however, they may appear at early times and vary in intensity. The neuroradiological exams are used for the diagnosis of BS, such as computed tomography, magnetic resonance imaging (MRI) and MR proton spectroscopy, which may demonstrate cerebral and cerebellar atrophy. It is also possible to perform fundoscopy of the eye to detect abnormal pigmentation of the retina and optic atrophy. Conclusion: Due to the difficult diagnosis of BS, a disease of an evolutionary nature, the clinical picture of the patient and its evolution are taken into account. However, neuroradiological exams may contribute to justify some clinical appearances in patients with BS, and genetic molecular mutation analysis may also be performed to better elucidate the prognosis of the disease.

Keywords: Batten syndrome; Lipofuscinosis; Mutation.

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