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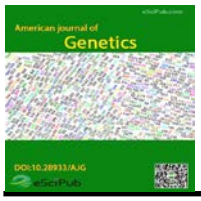

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Genetic Analysis of Clinical Manifestations of Friedreich Ataxia

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ABSTRACT

Introduction:Friedreich's ataxia (FA), an autosomal neurodegenerative disorder, conditions a destruction of nerve cells during the progression of the disease, affecting cardiac, bone and pancreatic cells. It has as main symptoms walking difficulty, progressing to changes in limb sensitivity, speech problems, atypical ocular movements, heart disease and diabetes. Objective: To carry out a review of the literature on Friedreich's ataxia and its association with genetic alterations and molecular diagnosis. Methodology: Articles published between 2010 and 2015 were pre-selected through the PubMed and SciELO databases, using the descriptor: ataxia. Analyzing the texts, 50 studies were initially identified. After reviewing the abstracts, considering the inclusion and exclusion criteria, 20 were analyzed, 3 were included in the review because they referred to the FA theme in the descriptors and abstracts. Results and Discussion: FA was identified through the genetic mapping, determinating chromosome 9 as the locus of malformation. This chromosome contains the frataxin gene, which encodes a frataxin protein, relating as mutations and causing abnormal repeats of glutamic acid (GAA). Homozygous for GAA represents 94% of patients with the classic form of the disease. The main clinical symptoms of FA are limb ataxia, cerebellar dysarthria, sensorimotor deficit in the lower limbs and pyramidal signs. Its diagnosis is made based on clinical investigations and confirmed from tests of molecular genetics. Thus, the genetic study associated with clinical investigation allows the prognosis, characterizing: a severity of the condition, its evolution and probability of being related to myocardiopathies. Conclusion: FA, an evolutionary disease, causes complete clinical changes after a few years from the beginning of the symptomatology, making early diagnosis difficult. Direct molecular diagnosis through the determination of GAA replicates becomes standard clinical examination, differential diagnosis and genetic counseling for individuals with cerebellar ataxias.

Keywords: Friedreich's ataxia; Epigenetics; Mutation

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