EDITORIAL

Editorial note on Machado-Joseph disease

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EDITORIAL NOTE

Machado–Joseph disease (MJD), also known as Machado–Joseph Azorean disease is a rare autosomal dominantly inherited neurodegenerative disease that causes progressive cerebellar ataxia which results in a lack of muscle control and coordination of the upper and lower extremities.

MJD is characterized by slowly progressive clumsiness in the arms and legs, a staggering lurching gait that can be mistaken for drunkenness, difficulty with speech and swallowing, impaired eye movements sometimes accompanied by double vision or bulging eyes, and lower limb spasticity. Some individuals develop dystonia (sustained muscle contractions that cause twisting of the body and limbs, repetitive movements, and abnormal postures) or symptoms similar to those of Parkinson's disease. Others may develop fasciculations (twitching) of the face or tongue, neuropathy, or problems with urination and the autonomic nervous system.

The disease is caused by a mutation in the *ATXN3* gene, which is located on chromosome 14q. In exon 10 the gene contains lengthy irregular repetitions of the code "CAG", producing a mutated protein called ataxin-3. (Normally, the number of copies is between 13 and 41. MJD is an autosomal dominant disease, meaning that if either parent gives the defective gene to a child, the child will show symptoms of the disease. Therefore, if one parent suffers from this disease and the other parent does not, there will be a 50% chance of their child inheriting the disease.

The pons (a structure located on the brain stem) is one of the areas affected by MJD. The striatum (a brain area connected to balance and movement) is also affected by this disease, which could explain both of the main motor problems cause by MJD: the tightening and twisting of the limb and the abrupt, irregular movements.

In affected cells, this protein builds up and assembles intranuclear inclusion bodies. These insoluble aggregates are hypothesized to interfere with the normal activity of the nucleus and induce the cell to degenerate and die.

MJD can be diagnosed by recognizing the symptoms of the disease and by taking a family history and through Genetic testing wherein we look at the number of CAG repeats within the coding region of the MJD/*ATXN3* gene on chromosome 14.

There is no cure for Machado-Joseph Disease. However, treatments are available for some symptoms. For example, spasticity can be reduced with antispasmodic drugs, such as baclofen. The Parkinsonian symptoms can be treated with levodopa therapy. Prism glasses can reduce diplopic symptoms, etc.

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