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My46 Trait Profile

Mucolipidosis 4

Other Names: Sialolipidosis

Mucolipidosis IV is a genetic condition most often characterized by movement and coordination problems, as well as progressive vision loss. It is caused by mutations in the MCOLN1 gene that makes a protein called mucolipin-1.

Characteristics of Mucolipidosis 4

Mucolipidosis type IV (ML IV) is a genetic condition that is associated with movement and coordination problems, progressive vision loss, and severe developmental delays/intellectual disability. The symptoms of ML IV typically appear in the first years of life. Children with the ML IV often have very limited speech and rarely learn to walk independently. The movement problems seen in ML IV are similar to those seen in cerebral palsy (e.g., low muscle tone, stiff muscles, abnormal posture, balance impairment). Problems with eating and swallowing are also commonly seen in this condition. Because of clouding and degeneration of parts of the eye, progressive vision loss is a common feature of ML IV. Individuals with ML IV often have low levels of digestive acids (e.g., gastrin) in the stomach, and high levels of gastrin in the blood. Anemia (i.e., low blood count) is another common feature seen in ML IV.

Diagnosis/Testing

Individuals with ML IV have changes or mutations in the MCOLN1 gene. This gene makes a protein called mucolipin-1. Mucolipin-1 appears to be important for the development of the retina and brain. This protein is also important for normal functioning of the stomach cells that produce digestive acids (e.g., gastrin). It is not entirely Mutations in the MCOLN1 gene either do not allow the protein to work properly or do not allow enough protein to be made. It is not entirely understood why and how the abnormal cilia cause the features seen in ML IV.

Management/Surveillance

Management of ML IV includes regular comprehensive eye exams, brain imaging, iron studies, and neurological evaluations to monitor disease. Intensive physical, occupational, speech, and vision therapies are also highly recommended.

Mode of inheritance

ML IV is inherited in an autosomal recessive pattern. This means that an individual has to inherit two MCOLN1 mutations (i.e., one from each parent) to be affected with Mucolipidosis IV. If both parents are carriers of a MCOLN1 mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with ML IV.

Risk to family members

Parents of a child with ML IV are carriers of ML IV. If a sibling of a child with ML IV is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of ML IV.

Special considerations

ML IV is estimated to occur in 1 in 40,000 people. About 70% of affected individuals have Ashkenazi Jewish

ancestry. Approximately 1 in 100 individuals with Ashkenazi Jewish ancestry is a carrier of ML IV.

Resources

Mucolipidosis IV Foundation
http://www.ml4.org
National MPS Society
http://www.mpssociety.org
Genetics Home Reference: Mucolipidosis type IV
http://ghr.nlm.nih.gov/condition/mucolipidosis-type-iv

References

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