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My46 Trait Profile

Mucopolysaccharidosis Type 4

Other Names: Morquio syndrome

Mucopolysaccharidosis type IV is a genetic condition characterized by the body's inability to break down complex sugar molecules called glycosaminoglycans. It is caused by mutations in either the GALNS gene that makes the N-acetylgalactosamine-6-sulfatase protein or the GLB1 gene that makes the beta-galactosidase protein.

Characteristics of Mucopolysaccharidosis Type 4

Individuals with mucopolysaccharidosis type IV (MPS IV) cannot properly break down complex sugar molecules called glycosaminoglycans (GAGs) because either the N-acetylgalactosamine-6-sulfatase enzyme or the beta-galactosidase enzyme is not working properly. Without enough enzyme, GAGs build up in various tissues of the body. As the GAGs accumulate, disease symptoms become debilitating and life threatening. There are two forms of MPS IV: MPS IVA and MPS IVB. The features seen are the same in both forms, but they each have a different genetic cause.

MPS IV is a progressive genetic disorder of bone and cartilage growth. The deformities of the bones and cartilage lead to pain and limited ability to move. Individuals with MPS IV have a short neck and bell-shaped chest, an abnormal curve to the spine, knock knees, flat feet, and short stature. Affected people may have a mild, but unique, facial appearance with a broad mouth, a short upturned nose, a large head, and teeth problems. Eye problems including clouding and glaucoma can contribute to vision loss. Heart problems and hearing loss are also seen with MPS IV. The symptoms of the condition usually begin between the ages of 1 and 3 years, and the life span for a person with MPS IV is usually about 20 years. Most individuals with MPS VI have normal intellectual abilities.

Diagnosis/Testing

A diagnosis of MPS IVA can be made by measuring the N-acetylgalactosamine-6-sulfatase enzyme activity from a blood sample. A person with MPS IVA often has deficient (low) enzyme activity. A diagnosis of MPS IVB can be made by measuring the beta-galactosidase enzyme activity from a blood sample. A person with MPS IVB often has deficient (low) enzyme activity.

A diagnosis of MPS IV disease can also be done by genetic testing for changes or mutations in the GALNS gene or the GLB1 gene. These genes make enzymes that are responsible for breaking down specific GAGs. Mutations in these genes prevent the enzymes from working properly, resulting in a build-up of specific GAGs in various organs of the body.

Management/Surveillance

Management of MPS IV often involves regular MRIs and x-rays, as well as routine clinical examinations by a healthcare professional familiar with MPS IV. Early therapies can result in fewer, more manageable symptoms and prevent complications. Currently, there are studies testing enzyme replacement treatments for MPS IVA.

Because most individuals with MPS IV have joint stiffness, limitation of motion, and bone problems, competitive and contact sports should be avoided. They should be as active as possible to maintain joint function and improve general health without increasing risk for injury. Physical and occupational therapies are recommended to achieve these goals.

Individuals with MPS IV are at an increased for anesthesia complications. Due to the accumulation of GAGs, there

is narrowing of the nasal passages and airways as well as enlargement of the tonsils, adenoids and tongue. This often makes it difficult to visualize the airway. It is important to select an anesthesiologist who has experience with difficult airways and it is recommended that the surgery or procedure be performed at an experienced hospital with an ICU in the event serious complications arise. Additionally, it is recommended that cardiac, respiratory, and upper-airway systems be evaluated before any procedure that requires sedation or anesthesia.

Mode of inheritance

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

Risk to family members

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

Special considerations

None

Resources

National MPS Society
http://www.mpssociety.org
Genetics Home Reference: Mucopolysaccharidosis type IV
http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-iv
The Carol Ann Foundation & The International Morquio Organization
http://www.morquio.com

References

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