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My46 Trait Profile

Mucopolysaccharidosis Type 6

Other Names: Maroteaux-Lamy syndrome, Arylsulfatase B deficiency

Mucopolysaccharidosis type VI is a genetic condition characterized by the body's inability to break down complex sugar molecules called glycosaminoglycans. It is caused by mutations in the ARSB gene that makes the arylsulfatase B protein.

Characteristics of Mucopolysaccharidosis Type 6

Individuals with mucopolysaccharidosis type VI (MPS VI) cannot properly break down complex sugar molecules called glycosaminoglycans (GAGs) because the enzyme, arylsulphatase B, 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.

MPS VI affects many systems of the body including the heart and blood vessels, the lungs, the brain and spinal cord, and the bones. Heart valve problems, hearing loss, increased mucous production, and airway narrowing are frequently seen in individuals with MPS VI. Lung infections and trouble breathing are commonly seen in this condition. The membrane that protects the brain and spinal cord (i.e., dura) may thicken due to the accumulation of the GAGs. This thickening can compress various nerves resulting in variety of symptoms including pain, vision problems, and compression of the spinal cord. Individuals with MPS VI typically have various bone deformities (e.g., kyphosis – excessive outward curving of the spine causing hunching of the back; contractures of the hands, elbows, shoulders, hips and knees). Individuals with MPS VI typically stop growing by the age of 8. As a result, they are short in stature, have joint stiffness and limited movement. Other characteristics of MPS VI include a unique facial appearance with thick lips, a low nasal bridge, a large head, and teeth problems. Individuals with MPS VI can also develop enlargement of the liver or spleen. The features seen in MPS VI, along with the age of onset and rate of progression is highly variable among affected individuals. Most individuals with MPS VI have normal intellectual abilities.

Diagnosis/Testing

A diagnosis of MPS VI can be made by measuring the arylsulphatase B enzyme activity from a blood sample. A person with MPS VI often has deficient (low) enzyme activity. A diagnosis of MPS VI can also be done by genetic testing for changes or mutations in the ARSB gene. This gene makes the arylsulphatase B enzyme that is responsible for breaking down specific GAGs. Mutations in the ARSB gene prevent the enzyme from working properly, resulting in a build-up of specific GAGs in various organs of the body.

Management/Surveillance

Individuals with MPS VI should be cared for by a team of healthcare providers including cardiologists, pulmonologists, ENTs, ophthalmologists, nutritionists, and physical and occupational therapists. Evaluations should be performed at least annually for monitoring the progression of the condition.

Enzyme replacement therapy (ERT) is a medical treatment available for MPS VI. ERT provides the arylsulphatase B enzyme that is missing in individuals with MPS VI through regular intravenous (IV) infusions. ERT is a lifelong treatment for MPS VI and is not a cure.

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

Risk to family members

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

Special considerations

None

Resources

National MPS Society
http://www.mpssociety.org
Genetics Home Reference: Mucopolysaccharidosis type VI
http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-vi
Maroteaux-Lamy syndrome Resource Center
http://www.maroteaux-lamy.com/en/hcp/index.aspx

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