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My46 Trait Profile

Mucopolysaccharidosis Type 2

Other Names: Hunter syndrome

Mucopolysaccharidosis type II 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 IDS gene that makes the iduronate-2-sulfatase protein.

Characteristics of Mucopolysaccharidosis Type 2

Individuals with mucopolysaccharidosis type I (MPS II) cannot properly break down complex sugar molecules called glycosaminoglycans (GAGs) because the enzyme, iduronate-2-sulfatase, 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.

Disease severity and onset of symptoms seen in MPS I are highly variable and affect many different parts of the body. The most common features of MPSII include umbilical and inguinal hernias, skeletal abnormalities, recurrent and persistent upper respiratory tract infections, distinct facial features, stiff joints, hydrocephalus, carpal tunnel syndrome, snoring, sleep apnea, hearing loss, enlarged spleen and liver, and heart problems.

Diagnosis/Testing

A diagnosis of MPS II can be made by measuring the iduronate-2-sulfatase enzyme activity from a blood sample. A person with MPS II often has deficient (low) enzyme activity. A diagnosis of MPS II can also be done by genetic testing for changes or mutations in the IDS gene. This gene makes the iduronate-2-sulfatase enzyme that is responsible for breaking down specific GAGs. Mutations in the IDS gene prevent the enzyme from working properly, resulting in a build-up of specific GAGs in various organs of the body.

Management/Surveillance

Management of MPS II focuses on improving quality of life, slowing down disease progression, and treating complications. Due to the progressive nature of MPS II, constant surveillance and evaluation of clinical signs and symptoms is recommended. This includes brain imaging, spine imaging, echocardiograms to monitor heart symptoms, hearing exams, pulmonary function testing, bone imaging and x-rays, and abdominal imaging to monitor liver and spleen size. There is no cure at this time for MPS II; however, early intervention and treatment may help prevent irreversible damage.

Enzyme replacement therapy (ERT) is a medical treatment available for MPS II. ERT provides the iduronate-2-sulfatase enzyme that is missing in individuals with MPS I through regular intravenous (IV infusions). ERT is a lifelong treatment for MPS I and is not a cure.

Because most individuals with MPS II 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 II 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 II is inherited in an X-linked recessive manner. This means that in females, both copies of the IDS gene (i.e., one on each X chromosome) must have a change or mutation, whereas in males, only one copy of the IDS gene must have a mutation to be affected. A female with a mutation in one copy of the IDS gene is said to be a "carrier" of MPS II, and is typically not affected.

Risk to family members

If a father is affected with MPS II, his daughters will be carriers of MPS II and his sons will be unaffected. If a mother is a carrier of MPS II, each daughter has a 1 in 2 chance (i.e., 50%) of being a carrier and each son has a 1 in 2 chance (i.e., 50%) of being affected with MPS II.

Special considerations

None

Resources

National MPS Society
http://www.mpssociety.org
HunterPatients
http://www.hunterpatients.com/
Genetics Home Reference: Mucopolysaccharidosis type II
http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-ii

References

Burton, BK. et al. (2012)."Diagnosing Hunter syndrome in pediatric practice: practical considerations and common pitfalls." European Journal of Pediatrics 171(4): 631-639.
Scarpa M. (Updated 22 February 2011). Mucopolysaccharidosis Type II. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1274/. Accessed [05/14/2013].
Walker, R. et al. (2013)."Anaesthesia and airway management in mucopolysaccharidosis." Journal of Inherited Metabolic Disease 36(2): 211-219.

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