Inheritest®
CARRIER SCREEN

Mucopolysaccharidosis type VI

What is mucopolysaccharidosis type VI?

Mucopolysaccharidosis type VI is an inherited metabolic disease affecting many tissues and organs.[1] Individuals with mucopolysaccharidosis type VI have defects in the enzyme arylsulfatase B, which breaks down large sugars known as glycosaminoglycans or mucopolysaccharides.[2] The symptoms of mucopolysaccharidosis type VI are due to the build-up of the large sugar molecules dermatan sulfate and chondroitin sulfate within lysosomes in cells.[2] Mucopolysaccharidosis type VI belongs to a group of diseases called lysosomal storage disorders and is also known as Maroteaux-Lamy syndrome.[1,2,3]

What are the symptoms of mucopolysaccharidosis type VI and what treatment is available?

Symptoms of mucopolysaccharidosis VI vary in severity and age at onset and are progressive. Affected individuals typically show symptoms in early childhood and are not intellectually impaired.[1,2] Signs and symptoms may include:[1,3]

- Skeletal and joint problems, including short stature
- Spinal cord compression
- Macrocephaly (large head) and hydrocephalus (fluid buildup in the brain)
- Distinctive facial features
- Airway narrowing, leading to respiratory infections and sleep apnea
- Heart valve abnormalities
- Hepatosplenomegaly (enlarged liver and spleen)
- Umbilical or groin hernias
- Corneal clouding, causing vision loss
- Frequent ear infections and hearing loss

There is no cure for mucopolysaccharidosis type VI. Treatment may include enzyme replacement therapy, supportive care, and hematopoietic stem cell transplantation.[3] The life expectancy of individuals with mucopolysaccharidosis type VI ranges from late childhood into adulthood, depending on the severity of symptoms.[1]

How is mucopolysaccharidosis type VI inherited?

Mucopolysaccharidosis type VI is an autosomal recessive disease caused by mutations in the *ARSB* gene.[1] An individual who inherits one copy of an *ARSB* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *ARSB* mutations, one from each parent, is expected to be affected with mucopolysaccharidosis type VI.

If both members of a couple are carriers of a mutation in the same gene, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for mucopolysaccharidosis type VI?

Mucopolysaccharidosis type VI can occur in individuals of all races and ethnicities. Its prevalence is estimated as 1 in 250,000 to 1 in 600,000 with a carrier frequency of 1 in 250 to 1 in 388.[1]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-vi

National MPS Society: http://mpssociety.org/mps/mps-vi/

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/maroteaux-lamy-syndrome/

References

1. Mucopolysaccharidosis type VI. *Genetics Home Reference.* Available at http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-vi. Accessed April 15, 2016.
2. *ARSB. Genetics Home Reference.* Available at http://ghr.nlm.nih.gov/gene/ARSB. Accessed April 15, 2016.
3. Valayannopoulos V, *et al.* Mucopolysaccharidosis VI. *Orphanet J Rare Dis* 2010 **5**:5