What is beta-mannosidosis?

Beta-mannosidosis is a rare inherited metabolic disease with variable severity and age of onset. Individuals with beta-mannosidosis have a deficiency of the beta-mannosidase enzyme, which helps to break down mannose-containing disaccharides (sugar molecules). The symptoms of beta-mannosidosis are due to the accumulation of disaccharides in the lysosomes. Beta-mannosidosis belongs to a group of diseases called lysosomal storage disorders.

What are the symptoms of beta-mannosidosis and what treatment is available?

The first onset of symptoms can range between infancy and adolescence. Symptoms can vary significantly in severity and may include:

- Intellectual disability
- Developmental delay
- Seizures
- Behavioral problems
- Increased risk of depression
- Respiratory and ear infections
- Hearing loss
- Speech impairment
- Swallowing difficulties
- Hypotonia (poor muscle tone)
- Peripheral nerve dysfunction
- Angiokeratomas (benign skin lesions)
- Skeletal abnormalities
- Distinctive facial features

Treatment is supportive and focuses on prevention of complications and management of symptoms.

How is beta-mannosidosis inherited?

Beta-mannosidosis is an autosomal recessive disease caused by mutations in the MANBA gene. An individual who inherits one copy of a MANBA gene mutation is a carrier. Carriers are not affected with beta-mannosidosis. An individual who inherits two mutations in the MANBA gene, one from each parent, is expected to be affected with beta-mannosidosis.

If both members of a couple are carriers of mutations in the same gene, the risk for 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 beta-mannosidosis?

Beta-mannosidosis is a rare condition and its prevalence is unknown.

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?


The International Advocate for Glycoprotein Storage Diseases (ISMRD): [http://www.ismrd.org/glycoprotein_diseases/beta_mannosidosis](http://www.ismrd.org/glycoprotein_diseases/beta_mannosidosis)

References