Beta-mannosidosis

Description

Beta-mannosidosis is a rare inherited disorder affecting the way certain sugar molecules are processed in the body.

Signs and symptoms of beta-mannosidosis vary widely in severity, and the age of onset ranges from infancy to adulthood. Almost all individuals with beta-mannosidosis experience intellectual disability, and some have delayed motor development and seizures. Affected individuals may be prone to depression or have behavioral problems such as hyperactivity, impulsivity or aggression. People with beta-mannosidosis are often extremely introverted.

People with beta-mannosidosis may experience an increased risk of respiratory and ear infections, hearing loss, speech impairment, swallowing difficulties, poor muscle tone (hypotonia), and reduced sensation or other nervous system abnormalities in the extremities (peripheral neuropathy). They may also exhibit distinctive facial features and clusters of enlarged blood vessels forming small, dark red spots on the skin (angiokeratomas).

Frequency

Beta-mannosidosis is believed to be a very rare disorder. It is difficult to determine the specific incidence of beta-mannosidosis, because people with mild or non-specific symptoms may never be diagnosed.

Causes

Variants (also known as mutations) in the *MANBA* gene cause beta-mannosidosis. The *MANBA* gene provides instructions for making the enzyme beta-mannosidase. This enzyme works in the lysosomes, which are compartments that digest and recycle materials in the cell. Within lysosomes, the enzyme helps break down complexes of sugar molecules (oligosaccharides) attached to proteins (glycoproteins). Beta-mannosidase is involved in the last step of this process, helping to break down complexes of two sugar molecules (disaccharides) containing a sugar molecule called mannose.

Variants in the *MANBA* gene interfere with the ability of the beta-mannosidase enzyme to perform its role in breaking down mannose-containing disaccharides. These

disaccharides gradually accumulate in the lysosomes and cause cells to malfunction, resulting in the signs and symptoms of beta-mannosidosis.

Learn more about the gene associated with Beta-mannosidosis

- MANBA

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have variants. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Beta-D-mannosidosis
- Beta-mannosidase deficiency
- Lysosomal beta A mannosidosis
- Lysosomal beta-mannosidase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Beta-D-mannosidosis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4048196/)

Genetic and Rare Diseases Information Center

- Beta-mannosidosis (https://rarediseases.info.nih.gov/diseases/869/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Beta-mannosidosis%22)

Catalog of Genes and Diseases from OMIM

- MANNOSIDOSIS, BETA A, LYSOSOMAL; MANSB (https://omim.org/entry/248510)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(beta-mannosidosis%5BTIAB%5D)+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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