Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Beta-Mannosidosis

Beta-Mannosidosis belongs to a group known as the Lysosomal Storage Disorders which occur as a result of specific enzyme deficiencies. These deficiencies mean that the body is unable to break down certain lipids or carbohydrates. If a person does not have enough of one of these enzymes, the body cannot break down the lipids or carbohydrates causing them to accumulate in the waste and recycling part of the cells called the lysosomes, preventing them from carrying out their normal function. In this disorder there is a deficiency of the beta-mannosidase enzyme which is needed to break down complexes of two sugar molecules containing a sugar molecule called mannose. The gene defect prevents the beta-mannosidase enzyme from breaking down these sugar molecules causing them to accumulate in the lysosomes, causing progressive damage to the cells leading to the symptoms and signs of this disorder.

This is a multisystem disorder, affecting may of the different organs and systems such as the brain, ears nose and throat, bones, muscle, joints and cartilage, skin, liver and spleen, chest, immune system, and the eyes. Growth may be delayed and intellectual and behavioural problems may be present. Beta-Mannosidosis is a very rare disorder and symptoms may vary considerably.

Synonyms

Alternative names for this condition are:

- Beta-D-Mannosidosis
- Beta-Mannosidase Deficiency
- Lysosomal Beta A Mannosidosis
- Lysosomal Beta-Mannosidase Deficiency

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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