Maple Syrup Urine Disease

Maple syrup urine disease, also called branched-chain ketoaciduria, is an autosomal recessive metabolic disorder caused by a deficiency of the alpha ketoacid dehydrogenase complex. This complex of enzymes is responsible for the degradation of branched chain amino acids including isoleucine, leucine, and valine. Defects in this enzyme cause the presence of sweet-smelling urine, similar to that of maple syrup leading to the name maple syrup urine disease. Infants with this metabolic disorder initially seem healthy at birth but if left untreated can suffer from seizures, CNS defects, and intellectual disability. Other nonspecific symptoms include poor feeding, vomiting, dehydration, lethargy and hypotonia. Treatment includes diet with minimal levels of branched chain amino acids necessary for metabolic functions.



PLAY PICMONIC

Pathophysiology

Defect in alpha ketoacid dehydrogenase
Broken Afro Key with Acidic-lemon Dehydrator
Maple syrup urine disease, also called branched-chain ketoaciduria, is a metabolic disorder caused by a deficiency of the alpha ketoacid dehydrogenase complex. This enzyme complex structure is analogous to alpha-ketoglutarate dehydrogenase and pyruvate dehydrogenase, requiring the same cofactors thiamine diphosphate, FAD, NAD, lipoate, and coenzyme A. Alpha ketoacid dehydrogenase is responsible for degradation of branched-chain amino acids.

Blocked degradation of branched chain amino acids
Branched Chains
Alpha ketoacid dehydrogenase is responsible for the degradation of branched-chain amino acids isoleucine, leucine, and valine.

Leucine
Lucy
Leucine is a branched-chain amino acid that accumulates in the blood and urine in maple syrup urine disease.

Isoleucine
Ice-Lucy
Isoleucine is a branched-chain amino acid that accumulates in the blood and urine in maple syrup urine disease.

Valine
Veil
Valine is a branched-chain amino acid that accumulates in the blood and urine in maple syrup urine disease.

Signs and Symptoms

Seizures
Caesar
A seizure is defined as a transient episode of abnormal, excessive neuronal activity. Seizures can be a symptom of maple syrup urine disease.

Intellectual Disability
Book Covered in Tar
Infants with this metabolic disorder initially seem healthy at birth but, if left untreated, can suffer from CNS defects and intellectual disability.

Sign up for FREE at picmonic.com