

MAPLE SYRUP URINE DISEASE

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Overview

Maple syrup urine disease is a rare but serious genetic disorder. The body is unable to process these amino acids, resulting in a buildup of harmful substances in the blood and urine that cannot be excreted.

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Causes

Maple Syrup Urine Disease is caused by a mutation in one gene. Mutations in these genes result in the absence or reduced activity of the human branched-chain alpha-ketoacid dehydrogenase complex (BCKAD) enzyme.

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Treatment

Treatment of MSUD consists primarily of lifelong therapy to maintain an acceptable diet and maintain normal metabolic status throughout life.

Diagnosis

Screening of a blood sample helps diagnose MSUD. Many infants with MSUD are found through newborn screening programs. A single blood sample can be screened for more than 40 different disorders.

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Symptoms

The main symptoms are a syrupy odor in urine, sweat, or earwax, lethargy, irritability, and not eating.

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