

## **Condition:** Maple syrup urine disease

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### **Inheritance:**

Autosomal recessive.

### **Genetic etiology:**

Mutation in genes (*BCKDHA*, *BCKDHB*, *DBT*) that encode subunits the enzyme complex branched chain amino acid dehydrogenase.

### **Frequency:**

Approximately 1/185,000.

### **Clinical features:**

MSUD results from deficiency of activity of branched chain amino acid dehydrogenase, leading to accumulation of leucine, isoleucine, and valine in infancy after the first feeding. Left untreated, severe neurological impairment ensues, leading to coma and death. Initial diagnosis is based on finding increased branched chain amino acids in plasma and ketones in the urine. There is a maple syrup odor in the urine, whence the name of the disorder.

### **Management:**

Dietary restriction of leucine and use of high calorie diet free of branched chain amino acids. Stress such as illness can precipitate metabolic decompensation, managed by treatment of the cause and reversing a state of protein breakdown by administration of fluids, sugars, and insulin.

### **Genetic counseling:**

Based on autosomal recessive inheritance. Newborn screening is widely practiced. Parents of affected child have 25% risk of recurrence; molecular genetic testing is available.