



Genetics Newborn Screening Program

Health Professional Fact Sheet

MAPLE SYRUP URINE DISEASE (MSUD)

Maple syrup urine disease (MSUD), a metabolic disorder, is caused by a deficiency in branched-chain ketoacid decarboxylation. The result is high body fluid (serum, urine, and spinal fluid) levels of leucine, isoleucine, valine, and their corresponding ketoacids. Lethargy, irritability, and vomiting progressing to coma and death occur in affected individuals if untreated. Screening in Connecticut was instituted in 1993.

GENETIC BASIS

- Autosomal recessive condition—Recurrence risk is 25%
- Prenatal diagnosis is possible

CT PREVALENCE 1:250,000 TO 400,000

CLINICAL FEATURES OF UNTREATED DISEASE

Severity

- Physical disabilities: Spastic quadriparesis, dystonic posturing, dysarthria, poor physical growth, seizures, central nervous system depression, coma, severe metabolic acidosis, hypoglycemia
- Developmental disabilities: irreversible mental retardation
- Mortality: Lethal usually within the first month of life

Symptomatic diagnosis

A symptomatic diagnosis is very possible and should be considered in any infant with severe acidosis in the first 10 days of life. Initial symptoms are poor feeding and marked lethargy along with a characteristic odor of the urine.

Variants

There are three variant forms of MSUD, intermediate, intermittent, and thiamine-response form. All three are associated with deficient decarboxylation of all three branched-chain ketoacids. Physical disabilities range from occasional developmental delay to mental retardation and ataxia.

CLINICAL OUTCOME WITH SCREENING AND TREATMENT

Mortality

Death within the first 2 weeks is not uncommon.

Clinical Disability

Treated patients frequently have irreversible retardation.

Variability

The outcomes of treated patients are only available for the classic form. Additional data are needed to evaluate the outcome of genetic variants detected by newborn screening programs.

Interventions

Dietary restriction of branched-chain amino acids requires frequent monitoring that must be continued indefinitely. Metabolic formula is available, and intake of branched-chain amino acids must be individually titrated. A nutritionist and metabolic specialists must coordinate therapy.

SCREENING FOR MSUD

Laboratory tests

- The test for elevated blood leucine content on filter paper blood specimen utilizing Tandem Mass Spectrometry.

Abnormal Test Results—Leucine levels \geq 4 mg/ml

- The primary care provider is notified by the DPH, Genetic NBS Tracking Unit nurse consultant.
- A referral is made by the DPH, Genetic NBS Tracking Unit nurse consultant to one of the Genetic Regional Treatment Centers.
- The primary care provider will be advised by the DPH, Genetic NBS Tracking Unit nurse consultant to contact the Genetic Regional Treatment Center to make a prompt referral and arrange for confirmation testing and evaluation.

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