

Condition Description

This inborn error of metabolism results in abnormally high levels of amino acids (leucine, isoleucine, valine). Without treatment central nervous system symptoms of lethargy, poor feeding, motor weakness, irritability progressing to seizures, coma and possibly death may occur shortly after birth. This disorder has a higher frequency of occurrence in the Mennonite population.

Range of Outcomes

With newborn screening most infants with MSUD are found pre-symptomatically and treated immediately with dietary restriction of amino acids leucine, isoleucine, valine, supplements and other therapies. Even with treatment outcomes are not perfect. Stress of acute illness may provoke a metabolic crisis requiring immediate and aggressive medical care to maintain health and avoid neurological sequelae. Factors which affect IQ are age of diagnosis, long-term metabolic control, fewer hospitalizations. In a cohort of 150 patients with classic MSUD 1/3 had IQ 70-90. 2/3 were over IQ 90. Cerebellar dysfunction was consistently documented by looking at verbal versus performance test results; verbal scores were higher than performance scores most of the time. Over half of early treated infants/children have short attention span and problems learning even with normal IQ. Literature has limited outcome data related to development and educational needs. An aggressive medical plan to respond to acute illness and injury is essential.

The outcomes for these infants depends on the ability of families to follow the restrictive dietary needs for infant, the infants response to therapy as well as the number, frequency and duration of hospitalizations.

Common Complications

Even with treatment, some children still develop swelling of the brain or have episodes of metabolic crisis. Children who have repeated metabolic crises may develop permanent brain damage. This can cause lifelong learning problems, intellectual disabilities or spasticity.

Inpatient treatment is essential during acute illness or injury because metabolic crisis at these times can be life-threatening. Brain edema, a common potential complication of metabolic decompensation, requires immediate therapy in an intensive care setting. (Strauss)

These infants may need frequent hospitalization. Infants need monitoring and evaluation to promptly respond to delays in the developmental domains. Medical plan for acute illness and injury and special dietary accommodations will always be essential components of a plan.

Side Effects of Treatment

None described.

For More Information

This fact sheet was developed for use in determining eligibility for early intervention services only. For more complete information, the following resources might be useful:

MedlinePlus

MedlinePlus brings together authoritative information from NLM, the National Institutes of Health (NIH), and other government agencies and health-related organizations. Preformulated MEDLINE searches also give access to medical journals.

<http://medlineplus.gov/>



Minnesota Children with Special Health Needs (MCSHN)
85 E. 7th Place, Suite 220
P.O. Box 64882
St. Paul, MN 55164
1-800-728-5420 or (651) 201-3650
www.health.state.mn.us/mcshn

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Agency for Healthcare Research and Quality

Under its Evidence-based Practice Program, the Agency for Healthcare Research and Quality (AHRQ) is developing scientific information for other agencies and organizations on which to base clinical guidelines, performance measures, and other quality improvement tools. Contractor institutions review all relevant scientific literature on assigned clinical care topics and produce evidence reports and technology assessments, conduct research on methodologies and the effectiveness of their implementation, and participate in technical assistance activities.

<http://www.ahrq.gov/clinic/epcsums/lbwdissum.htm>

E-Medicine

eMedicine's clinical knowledge base contains peer reviewed articles on a number of different health conditions. There is a pediatric section available.

<http://www.emedicine.com/>

References

Strauss, K., et al. "Maple Syrup Urine Disease", Geneclinics.org. updated January 2006.

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Morton DH, Strauss KA, Robinson DL, et al: Diagnosis and treatment of maple syrup disease: a study of 36 patients. *Pediatrics* 2002 Jun; 109(6): 999-1008

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