www.msud-support.org

Maple Syrup Urine Disease Fact Sheet

What is MSUD?

Maple syrup urine disease (MSUD) is a rare genetic disorder characterized by a deficiency of the enzyme needed to break down certain amino acids, the building blocks of protein. The result of this metabolic failure is a buildup of these amino acids and their byproducts to toxic levels. It received its name due to an odor similar to maple syrup which is detected in urine and earwax.

Individuals with MSUD cannot survive without the medical formula as they must restrict all sources of dietary protein including meat, fish, dairy, nuts, beans, and grains (breads and cereals).

Who is at risk for MSUD?

MSUD occurs in all ethnic groups with an estimated incidence of 1 in 185,000 live births, but certain populations are affected at a much higher frequency. The Old Order Mennonite population has an estimated incidence of about 1 in 380 newborns, and the Ashkenazi Jewish population has an estimated incidence of 1 in 26,000 newborns due to founder effects.

How is MSUD Manifested?

Babies born with MSUD experience neurological dysfunction within the first few days of life characterized initially by lethargy, irritability and poor feeding. This is soon followed by convulsions and deepening coma. If untreated, progressive brain damage is inevitable and death ensues usually within weeks or months. Cognitive disabilities such as ADD, anxiety and depression are common due to the effects on the brain.

How is MSUD Diagnosed?

MSUD is diagnosed through the newborn screening panel days after birth in all 50 US states.

How is MSUD Treated?

The only treatment for this disease is strict adherence to a specialized diet. This requires a specific medical food (formula) which provides all of the amino acids needed for growth except those that cannot be metabolized. These must be provided in specific amounts determined for each individual by frequent blood tests. Failure to provide adequate amounts results in impaired growth and breakdown of body tissue, while excessive intake results in toxic levels in the blood and brain resulting in neurological damage.

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Even with treatment, those with MSUD remain at high risk for developing episodes of acute illness (metabolic crises) which may be triggered by infection, injury, failure to eat (fasting) or even by psychological stress. During these episodes there is a rapid, sudden spike in amino acid levels necessitating immediate medical intervention.

Why is the Medical Nutritional Equity Act (MNEA) important to MSUD families?

Medical treatment and medical foods are expensive. Insurance coverage for treatment and medical food varies widely between states and within states where legislation does not require coverage. Without adequate access, hospitalizations will increase in frequency and brain damage will occur resulting in an inability to care for oneself.