MSUD
FAMILY SUPPORT GROUP

Introduction

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).

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.

Concerns:

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.

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 occurs in all ethnic groups with an incidence of 1 in 180,000 live births, with an estimated 1,000 affected in the US. It received its name due to an odor similar to maple syrup which is detected in urine and earwax.

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 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.

Why the Medical Foods and Formulas Access Act of 2025 is a priority:

Medical treatment as well as medical foods, formula and supplements 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. Death has occurred in states where Medicaid does not cover formula for adults.

Our group has joined with several other stakeholders to form a coalition called the Patients & Providers of Medical Nutrition Equity. The passage of the Medical Foods and Formulas Access Act of 2025 is critical to the health and well-being of our constituents. Please sign on as a co-sponsor when it is reintroduced in Congress.