



MAPLE SYRUP URINE DISEASE (MSUD) CONSUMER SUMMARY

SERN/GMDI Nutrition Management Guidelines

First Edition

Nutrition Guidelines for Individuals with MSUD^{1,2,3}

November 2018

This information is intended for individuals with Maple Syrup Urine Disease (MSUD) and/or their caregivers (hereafter referred to as “you/your”). This summary includes current management recommendations and suggests topics that you can discuss with your metabolic team to help identify a plan that is best for you.

- The goal of MSUD nutrition management is to control the concentration of the branched-chain amino acids (BCAA) in your blood in order to optimize your growth and development, and to prevent metabolic decompensation. Leucine (LEU) levels should be maintained between 75-200 µmol/L for infants and children aged five years or younger, and between 75-300 µmol/L for individuals over the age of five years. Isoleucine (ILE) and valine (VAL) levels should be maintained between 200-400 µmol/L (or slightly above the normal ranges) in individuals with MSUD.
- BCAA concentrations (LEU, ILE, VAL) are maintained by a combination of restricting protein in the diet and by consuming a special medical food (formula). Individualized recommendations for protein restriction and medical foods will depend on your age, weight and height, blood BCAA control and whether you are sick or healthy.
- For infants with MSUD, breast milk may be acceptable as part of the nutrition management plan if there is frequent monitoring of the infant’s growth, health and BCAA concentrations.
- During times of serious illness, more intensive nutrition management is required to prevent or reverse metabolic decompensation. This may include extra calories, medical food, further protein restriction and fluids. Other medical treatments, such as dialysis, also may be required.
- Work with your metabolic clinic to always have a sick-day plan and an emergency letter to use when seeking urgent medical care.
- Thiamin supplementation may increase the amount of protein allowed in the diet in some individuals with MSUD. Your clinic may try a thiamin supplement to determine whether or not more thiamin is helpful for you.
- If you are a female with MSUD, obtain information from your metabolic team about the impact of pregnancy on MSUD management and the changes to your diet, medications, supplements and monitoring that may be needed if you become pregnant.
- If you are a female with MSUD, and wish to breastfeed your infant, your clinic will closely monitor your nutrient intake, health status and BCAA concentrations, as well as your infant’s growth and development.
- Liver transplantation is a treatment option for individuals with MSUD. It is optimal to have your BCAA concentrations in control before transplantation. After a successful transplantation, you will no longer be required to follow a diet with a protein-restriction and medical food, although your clinic may assist in transition to a regular diet and continue to monitor your growth and nutrient status.

This document is not meant to substitute for the medical advice provided by your doctor.

¹ For children, adolescents, and adults with MSUD, or their caregivers

² Based on the 2017 Nutrition Management Guidelines for Maple Syrup Urine Disease (MSUD) by Genetic Metabolic Dietitians International (GMDI) / Southeast Regional Genetics Network (SERN): https://southeastgenetics.org/ngp/guidelines_msud.php

³ The Management Guidelines Advisory Committee used the nationally standardized condition abbreviation of MSUD; curated by the US National Library of Medicine for this and related guideline products: <https://newbornscreeningcodes.nlm.nih.gov/>

