



MAPLE SYRUP URINE DISEASE (MSUD) FREQUENTLY ASKED QUESTIONS

SERN/GMDI Nutrition Management Guidelines

First Edition

F.A.Q. About Nutrition Management for Individuals with MSUD^{1,2,3}

January 2020

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NUTRIENT INTAKE	
<i>Can an infant with MSUD be breastfed?</i>	A limited amount of breast milk can be included in the dietary management of infants with MSUD if there is frequent monitoring of the baby's health, growth, development, and Branch Chain Amino Acid (BCAA) blood concentrations (Rec. 1.7).
<i>How much Leucine (LEU) or protein can I eat?</i>	The amount depends on your individual LEU tolerance, age, weight and needs for growth (in children) or health maintenance (in adults). Your metabolic clinic will use your lab results and your health status to recommend LEU or protein intake goals (Rec 2.4).
<i>Do I need an MSUD medical food (formula)?</i>	Most individuals with MSUD need medical food because your LEU restriction does not allow enough protein from food for appropriate growth and health. Your protein intake will usually be provided by a combination of food and the medical food recommended by your metabolic clinic.
<i>How do I know if I am getting the right number of calories?</i>	Your calorie needs are based on age, weight, activity level and growth. Your clinic will make recommendations that are appropriate for you (Rec 2.4).
MONITORING	
<i>What are the goals for my leucine, isoleucine, and valine blood concentrations?</i>	Blood LEU concentration 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 (Rec 2.1). Isoleucine (ILE) and valine (VAL) concentration should be maintained between 200-400 µmol/L (or slightly above the normal ranges) in individuals with MSUD (Rec 2.2). Blood BCAA concentrations should be maintained within the recommended ranges throughout life (Rec 2.3).
<i>How often should monitoring of my MSUD be done?</i>	Your metabolic clinic will recommend how often blood BCAA testing should be done (based on past lab results and any adjustments needed for growth or activity level) and how often you should come to clinic. They may also recommend additional testing to monitor your nutritional needs.
ILLNESS	
<i>What should I do if I become ill?</i>	Call your metabolic physician. If urgent care is required, take your emergency letter with you.
<i>How does nutrition management change during illness?</i>	During 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 (Rec. 1.1). Other medical treatments, such as dialysis, may also be required. Mild illnesses may be managed according to patient-specific sick day protocols provided by your metabolic clinic (Rec 1.8).

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

¹ For the child, teenager, or adult living with MUSD and their caregivers.

² Based on the Nutrition Management Guidelines for Maple Syrup Urine Disease (MSUD) by GMDI/SERC 2018: https://southeastgenetics.org/ngp/guidelines_msud.php

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

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LIVER TRANSPLANT	
<i>How does treatment for MSUD change after a liver transplant?</i>	After transplant, a person with MSUD may be able to increase their protein intake, without the use of medical food, to the recommended amount for someone their age without MSUD (Rec 5.4). Your clinic may assist you in your transition to a regular diet, and may continue to monitor your growth and nutrient status (Rec 5.5).
<i>How much LEU or protein can I eat?</i>	The amount depends on your individual LEU tolerance, age, weight and needs for growth (in children) or health maintenance (in adults). Your metabolic clinic will use your lab results and your health status to recommend LEU or protein intake goals (Rec 2.4).
PREGNANCY	
<i>How does pregnancy change my MSUD management?</i>	Your metabolic clinic will adjust your treatment recommendations to provide LEU control (Rec 4.2) and meet the additional nutritional needs of pregnancy (Rec 4.3). Your recommendations for dietary intake will be adjusted to maintain blood LEU concentrations in the 100-300µM range and VAL and ILE in the upper range of normal (200-400µM) throughout pregnancy. More frequent monitoring and clinic visits will be needed throughout pregnancy and after the baby is born (Rec 4.4).
<i>Can a woman with MSUD breastfeed her baby?</i>	A woman with MSUD may be able to breastfeed her infant if she works closely with her metabolic clinic to monitor her nutrient intake, health and laboratory test results, and her infant's growth and development (Rec 4.6).
SUPPLEMENT	
<i>Do I need to take a thiamin supplement?</i>	Except for individuals with certain genetic variations, a thiamin challenge is recommended to determine if extra thiamin is helpful for you (Rec. 3.1, 3.2, 3.3). If you are responsive to thiamin and continue to take it as a supplement, you may be able to increase your intake of protein under the direction of your clinic staff.

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