



At a Glance Maple Syrup Urine Disease (MSUD)

Deficient enzyme: Branched-chain keto acid dehydrogenase enzyme complex

Toxic Metabolite: Leucine and its keto acid (2-oxo-isocaproic acid)

Restricted Amino Acid: Branched-chain amino acids (BCAA)

Clinical presentation, in untreated patients:

Classic: neonatal onset, poor feeding, lethargy, altered tone, ketoacidosis, seizures, developmental delays

Intermediate: failure to thrive, ketoacidosis, developmental delays, classic symptoms during catabolic illness

Intermittent: normal development, episodic ataxia, ketoacidosis

***Goal Treatment Range:** Maintain plasma BCAA as close to normal as possible. Acceptable ranges:

Leucine: 100-300 $\mu\text{mol/L}$

Isoleucine: 100-300 $\mu\text{mol/L}$

Valine: 200-400 $\mu\text{mol/L}$

* van Calcar, S. Nutrition Management of Maple Syrup Urine Disease. In LE Bernstein, F Rohr, S van Calcar (Eds.) *Nutrition Management of Inherited Metabolic Diseases (2nd Edition)*. Springer: 2021

Nutrient Needs by Age*

Age	Leucine mg/kg/d	Isoleucine mg/kg/d	Valine mg/kg/d	Intact Protein [†] g/kg/d	Total Protein g/kg/d	Energy kcal/kg/d
0-6 months	40-100	36-100	40-95	1.0-1.6	2.5-3.5	95-145
7-12 months	40-75	30-70	30-80	0.8-1.4	2.5-3.0	80-135
1-3 years	40-70	20-70	30-70	0.6-1.2	1.5-2.5	80-130
4-8 years	35-65	20-30	30-50	0.4-0.9	1.3-2.0	50-120
9-13 years	30-60	20-30	25-40	5.0-8.0 g/day	1.2-1.8	40-90
14-18 years	15-50	10-30	15-30	5.0-8.0 g/day	1.2-1.8	35-70
19+ years	15-50	10-30	15-30	5.0-8.0 g/day	1.1-1.7	35-45

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[†]SERN/GMDI MSUD Management Guidelines; <https://southeastgenetics.org/ngp/guidelines.php>

Starting a MSUD Diet*

- Determine goals for Leucine (mg), Intact Protein (g), Total Protein (g), Energy (kcal)
- if estimating mg of leucine from grams of protein: 60 mg leucine is ~1 g protein
- Calculate amount of intact protein source (breast milk, infant formula, food) needed to meet Leu goal.
- Calculate amount of medical food needed in addition to the intact protein source to meet total protein goal.
- Calculate energy intake from intact protein and medical food sources to ensure total calorie needs are met.

Diet During Illness*

In consultation with the medical team, if the patient's plasma leucine is significantly elevated:

- Reduce intact protein by 50-100%, depending on the leucine levels and severity of illness, until plasma leucine is in the treatment range. Withholding all intact protein for extended periods may lead to catabolism.
- Increase medical food and non-protein energy sources to support anabolism.
- Add L-isoleucine and L-valine supplements (20-120 mg/kg/d of each) to maintain plasma isoleucine and valine higher than the normal treatment range. Goal is 400-800 $\mu\text{mol/L}$.

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	Abbott abbottnutrition.com	Mead Johnson hcp.meadjohnson.com	Nutricia NutriciaMetabolics.com	VitaFlo www.vitafloUSA.com
Infant (0-1 yr)	Ketonex®-1	BCAD 1	MSUD Anamix® Early Years	
Toddler & Young Children	Ketonex®-1 Ketonex®-2	BCAD 1 BCAD 2	MSUD Anamix® Early Years Complex Junior MSD Drink Mix Complex Essential MSD Drink Mix Complex MSD Amino Acid Blend	MSUD gel™ MSUD express® 15 MSUD cooler® 15
Older Children & Adults	Ketonex®-2	BCAD 2	Complex Essential MSD Drink Mix Complex MSD Amino Acid Blend MSUD Maxamum® MSUD Lophlex® LQ	MSUD express® 15 MSUD cooler® 15

Nutrition Supplementation

Thiamine*

-Trial of 100-1,000 mg/d to determine responsiveness (only effective in variant forms of MSUD)

L-isoleucine and L- valine*

- given to maintain plasma ILE and VAL in treatment range (dose varies)
-used during metabolic crisis to decrease plasma leucine (see Diet During Illness)

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Laboratory Monitoring

Plasma Leucine^{1,2}

Plasma Amino Acids^{1,2}

Ketones^{1,2}

Prealbumin³

Albumin³

Ferritin³

CBC³

¹ Daily until stable, weekly to twice weekly until 6 months old

² Monthly after 24 months of age

³ Every 6 months

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