At a Glance
Methylmalonic/Propionic Acidemia

Deficient enzyme: MMA-methylmalonyl-CoA mutase (mut^0 or mut^-)
PROP- propionyl-CoA carboxylase

Cofactor: MMA- Adenosylcobalamin (Vitamin B_{12})
PROP- Biotin

Toxic Metabolite: MMA- Methylmalonic Acid
PROP-Propionic Acid

Restricted Amino Acids: Valine, Isoleucine, Methionine, Threonine

Clinical presentation in untreated patients: acute: poor feeding, vomiting, lethargy, tachypnea, acidosis, respiratory distress, coma; longer-term: neurologic complications, optic atrophy, renal dysfunction (MMA), cardiomyopathy (PROP)

***Goal Treatment Range**: Plasma amino acids- maintain within normal range

Nutrient Needs by Age*

Age	Intact Protein g/kg/d	Total Protein g/kg/d	Energy kcal/kg/d
0-3 mo	0.9 – 1.5	1.5 – 1.8	72 - 109
3-6 mo	0.9 – 1.5	1.5 – 1.8	72 - 109
7-12 mo	0.7 – 1.2	1.2 – 1.4	64 - 97
1-3 yrs	0.6 – 1.05	1.0 – 1.2	66 - 99
4-8 yrs	0.57 – 0.95	0.95 – 1.1	56 - 88

*SERN/GMDI PROP Nutrition Management Guidelines; https://southeastgenetics.org/ngp/guidelines.php

Starting a Diet

1. Determine goals for Intact Protein (g), Total Protein (g), Energy (kcal)
2. Calculate amount of intact protein source (breast milk, infant formula, food) needed to meet Intact Protein (g) goal.
3. Calculate amount of medical food required to provide remaining protein to meet total protein goal.
4. Calculate energy intake from intact protein and medical food sources to ensure total calorie needs are met.

Methylmalonic/Propionic Acidemia

	Abbott abbottnutrition.com	Mead Johnson hcp.meadjohnson.com	Nutricia NutriciaMetabolics.com	Vitaflo www.vitafloUSA.com
Infant (0-1 yr)	Propimex®-1	OA 1	MMA/PA Anamix® Early Years	
Toddler & Young Children	Propimex®-1 Propimex®-2	OA 1 OA 2	MMA/PA Anamix® Early Years MMA/PA Anamix® Next	MMA/PA gel™ MMA/PA express®15 MMA/PA cooler®15
Older Children & Adults	Propimex®-2	OA 2	MMA/PA Anamix® Next XMTVI Maxamum®	MMA/PA express®15 MMA/PA cooler®15

Nutrition Supplementation (dose may vary based on blood laboratory results)

L-Carnitine: 100-300 mg/kg/d; divided two to four times per day

*MMA- Hydroxycobalamin: 1.0-2.0 mg daily to weekly for those who are vitamin B_{12} responsive (these patients may need little to no dietary restriction)

To determine responsiveness: 1.0 mg (IM or IV) hydroxycobalamin x 5 days; reduction in MMA levels of ≥50% indicates responsiveness

**PROP- Biotin: 5-40 mg/d to determine responsiveness and if a non-responder, stop biotin

*Sowa, M. Nutritional Management of Propionic and Methylmalonic Acidemia. In LE Bernstein, F Rohr, S van Calcar (Eds.) Nutritional Management of Inherited Metabolic Diseases (2nd Edition). Springer: 2021
**Jurecki E et al. Nutrition management guideline for propionic acidemia: An evidence- and consensus-based approach. Mol Genet Metab. 2019;126(4):341-54

Medical Therapy

Carbaglu (carglumic acid) (www.recordati.com)
Maintenance dose for chronic hyperammonemia (pediatric and adults) 10-100 mg/kg/d

Laboratory Monitoring*

Plasma Amino Acids[1]
Serum Methylmalonic Acid[1]
Carnitine (Free and Acyl)[1]
Ketones[1] (continue monthly in PROP

CBC, Albumin[2]
Prealbumin[2]
Propionic Acid[3]

Urine organic acids[3]
25-OH Vitamin D[4]
Folate, ferritin, B_{12}, B_6, zinc, selenium
Complete Metabolic Panel (CMP)[4]

[1] Monthly in infancy; every 3-6 months thereafter
[2] Every 6 months in infancy; annually thereafter
[3] Every 6 months in infancy; then annually as indicated
[4] Annually

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