

Methylmalonic Acidemia (mutase)

Ages 6-12 months

Clinic Visits:

- At least every 3 months

Laboratory studies:

- Monitoring of diet
 - Plasma amino acid levels and methylmalonic acid level at least monthly
- Yearly
 - Comprehensive metabolic panel
 - Urine analysis for protein
 - Blood pressure check
 - Serum carnitine
 - Plasma amino acids
 - Urine organic acids
 - Quantitative urine/plasma methylmalonic level
 - Prealbumin /albumin
 - Plasma ferritin, transferrin, or iron studies
 - Amylase and Lipase
 - Possible B-type Natriuretic peptide (heart)
 - Possible CBC, hemoglobin, and hematocrit
 - Consider folate and Vitamin B12
 - Consider other nutritional testing depending on formula (Zinc, selenium, vitamin D, essential fatty acids, and lipid profile).

During illness:

- Call Metabolic Clinic to alert them immediately if your child is sick
 - Poor feeding, fever, vomiting, lethargy
- Give your child glucose gel or polymer solution if needed (or instructed to do so)
- If your child is not able to keep down glucose containing fluids, you will need to take him or her to the emergency room for IV 10% dextrose (glucose)
- Be sure to bring Emergency Room letter with you to the hospital
- Labs during illness include: comprehensive metabolic panel, phosphate, CBC, urine analysis for ketones, amylase/lipase (risk of pancreatitis), possible ammonia, possible B-type Natriuretic peptide (heart), possible plasma amino acids, and possible methylmalonic acid
- IV carnitine will be given to your child
- Please alert the Metabolic Clinic if your child needs surgery
- Consider Medic Alert bracelet

Evaluations:

- Possible neurology or Rehab evaluation if your child shows signs of brain involvement

Topics:

- Biochemistry and genetics of methylmalonic acidemia
- Complications
 - Bone marrow and immune suppression
 - Pancreatitis
 - Kidney disease

Diet/management

- Propionogenic amino acid restricted diet
- Importance of medical formula
- Introduction of low protein foods
- Possible B12 injections
- Carnitine
- Possible use of antibiotics to reduce gut flora
- Possible transplant of liver and/or kidney
- Avoid fasting
- Management during illness

Please make sure your child has clinic visits and levels done on time. Call the IMD clinic at _____ to make an appointment, schedule a blood draw, or with any other questions. If you urgently need to reach a Metabolic physician, call _____ and ask to page the Metabolic physician on call.

Thank You,
Your Metabolic Team