Methylmalonic Acidemia (mutase)
1-18 years

Clinic Visits:

- ☑ At least every 6 months

Laboratory studies:

- ☑ Monitoring of diet
  - 1-6 years: plasma amino acids and methylmalonic acid levels at least every 3 months
  - 6-18 years: plasma amino acids and methylmalonic acid levels at least every 6 months
- ☑ 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
  - Possible folate and Vitamin B12
  - Consider other nutritional testing depending on formula (Zinc, selenium, vitamin D, essential fatty acids, and lipid profile).
  - Possible 24 hour urine creatinine clearance and protein at 6 years old and yearly thereafter.

During illness:

- ☑ Call Metabolic Clinic to alert them that 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
- ☑ Possible cardiac monitoring while in the hospital
- ☑ Please alert the Metabolic Clinic if your child needs surgery
- ☑ **Wear Medic Alert Bracelet!!!**

Evaluations:

- ☑ Yearly developmental questionnaires to be completed by parents
- ☑ Developmental evaluation at 3 and 6 years old

- ☑ Neuropsychological evaluation at 9 years old
- ☑ DEXA scan (bone health) at 9 and 18 years old
- ☑ Possible neurology or rehab evaluation if needed
- ☑ Possible renal evaluation (kidneys) if needed

Topics:

- ☑ Review of biochemistry and genetics of methylmalonic acidemia
- ☑ Complications
    - o Bone marrow and immune suppression
    - o Pancreatitis
    - o Kidney disease
- ☑ Diet/management
    - o Propiongenic amino acid restricted diet
    - o Importance of medical formula
    - o Use of low protein foods
    - o Possible B12 injections
    - o Carnitine
    - o Possible use of antibiotics to reduce gut flora
    - o Possible transplant of liver and/or kidney
    - o Avoid fasting
    - o 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