

Methylmalonic Acidemia (mutase) >18 years

Clinic Visits:

- At least yearly

Laboratory studies:

- Monitoring of diet
 - o plasma amino acids and methylmalonic acid levels at least yearly
- Yearly
 - o Comprehensive metabolic panel
 - o Urine analysis for protein
 - o Blood pressure check
 - o Serum carnitine
 - o Plasma amino acids
 - o Urine organic acids
 - o Quantitative urine/plasma methylmalonic level
 - o Prealbumin /albumin
 - o Plasma ferritin, transferrin, or iron studies
 - o Amylase and Lipase
 - o Possible B-type Natriuretic peptide (heart)
 - o Possible CBC, hemoglobin, and hematocrit
 - o Consider folate and Vitamin B12
 - o Consider other nutritional testing depending on formula (Zinc, selenium, vitamin D, essential fatty acids, and lipid profile).
 - o Possible 24 hour urine creatinine clearance and protein

During illness:

- Call Metabolic Clinic to alert them that you are sick
 - o Poor feeding, fever, vomiting, lethargy
- Take glucose gel or polymer solution if needed (or instructed to do so)
- If you are not able to keep down glucose containing fluids, you will need to go 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 you
- Possible cardiac monitoring while in the hospital
- Please alert the Metabolic Clinic if you need surgery
- Wear Medic Alert Bracelet!!!**

Evaluations:

- DEXA scan (bone health) at 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
 - Bone marrow and immune suppression
 - Pancreatitis
 - Kidney disease
- ☑ Diet/management
 - Propionic amino acid restricted diet
 - Importance of medical formula
 - Use 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 you have 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