

# Methylmalonic Acidemia (mutase)

## Ages 0-6 months

### Clinic Visits:

- At least every 2 months

### Laboratory studies:

- At diagnosis
  - Urine organic acids
  - Plasma acylcarnitine profile
  - Serum amino acids
  - Comprehensive metabolic panel
  - Ammonia
  - CBC
  - Urine analysis (for ketones)
  - Serum B12 level
  - Plasma total homocysteine
  - Possible quantitative plasma methylmalonic acid
  - Possible complementation studies on cultured fibroblasts (skin biopsy)
  - Possible DNA studies of MMA genes
- Monitoring of diet
  - Plasma amino acid and methylmalonic acid levels at least monthly

### During illness:

- Call Metabolic Clinic to alert them immediately if your baby is sick
  - Poor feeding, fever, vomiting, and/or lethargy
- Give your baby glucose gel or polymer solution if needed (or instructed to do so)
- If your baby 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 baby
- Please alert the Metabolic Clinic if your baby needs surgery

### Evaluations:

- Possible neurology or Rehab evaluation if your baby shows signs of brain involvement
- Possible brain MRI
- Possible renal (kidney) evaluation

### 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
- 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 metabolic 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

