

Maple Syrup Urine Disease

Ages 0-6 months

Clinic Visits:

- Every 2 months

Laboratory studies:

- At diagnosis
 - Plasma amino acids
 - Possible urine organic acids
 - Basic metabolic panel
 - Possible osmolarity studies if your baby is showing symptoms
 - Possible molecular analysis of BCKAD gene
 - Possible skin biopsy (enzymatic studies)
- Monitoring of diet
 - Plasma amino acid levels weekly
 - Treatment range:
 - Leucine <500 µmol/L
 - Isoleucine >1000 µmol/L
 - Valine >100 µmol/L

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: basic metabolic panel, plasma amino acids, osmolarity, and amylase/lipase (risk of pancreatitis)
- Your baby is at great risk of brain edema which may require a head CT scan and possibly dialysis
- Please alert the Metabolic Clinic if your baby needs surgery

Evaluations:

- Possible brain MRI if your baby shows any neurological symptoms
- Possible liver evaluation to discuss transplant options

Topics:

- Biochemistry and genetics of MSUD
- Complications
 - Brain edema
 - Pancreatitis
- Diet/management
 - Leucine-, Isoleucine-, and Valine-restricted diet
 - Importance of medical formula

- Avoid fasting
- Management during illness
- Possible thiamine supplementation
- Possible iron supplementation

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.

Thank You,
Your Metabolic Team

