

Maple Syrup Urine Disease

Ages 2-18 years

Clinic Visits:

- At least every 6 months

Laboratory studies:

- Monitoring of diet
 - 2-3 years – plasma branched chain amino acid levels monthly
 - 3-18 years – plasma branched chain amino acid levels every 3 months
 - Treatment range:
 - Leucine <500 µmol/L
 - Isoleucine >1000 µmol/L
 - Valine >100 µmol/L
- Yearly:
 - Basic metabolic panel
 - Prealbumin /albumin
 - Ferritin, transferritin, or iron studies
 - Amylase and lipase
 - Possible CBC, hemoglobin, and hematocrit
 - Possible folate and vitamin B12
 - Possible nutritional labs (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, and/or 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: basic metabolic panel, plasma amino acids, osmolarity, and amylase/lipase (risk of pancreatitis)
- Your child is at great risk of brain edema which may require a head CT scan and possibly dialysis
- 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 age 3 and 6 years
- Neuropsychological evaluation at age 9 years
- Psychiatric screening at age 18 years
- DEXA scan (bone health) at 9 and 18 years
- Possible brain MRI if your child shows any neurological symptoms
- Possible liver evaluation to discuss transplant options

Topics:

- Review of biochemistry and genetics of MSUD
- Complications

- Brain edema
- Pancreatitis
- ☑ Diet/management
 - Leucine-, Isoleucine-, and Valine-restricted diet
 - Importance of medical formula
 - Introduction of low protein foods
 - 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