

# Maple Syrup Urine Disease

## Ages >18 years

### Clinic Visits:

- At least every 6 months

### Laboratory studies:

- Monitoring of diet
  - 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 if you are sick
  - Poor feeding, fever, vomiting, and/or lethargy
- Take glucose gel or polymer solution if needed (or instructed to do so)
- If you are unable 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)
- You are at great risk of brain edema which may require a head CT scan and possibly dialysis
- Please alert the Metabolic Clinic if you need surgery
- Wear Medic Alert bracelet!**

### Evaluations:

- Possible brain MRI if you show any neurological symptoms

### 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 you have 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