

Isovaleric Acidemia

>18 years

Clinic Visits:

- At least yearly

Laboratory studies:

- Monitoring of diet
 - plasma amino acid levels at least yearly
- Yearly
 - Comprehensive metabolic panel and phosphorous
 - Amylase and lipase (risk of pancreatitis)
 - Carnitine levels
 - Prealbumin/albumin
 - Ferritin, transferrin, or iron studies
 - Possible urine organic acids
 - Possible CBC, hemoglobin, and hematocrit
 - Possible folate and vitamin B12
 - Possible nutritional testing: zinc, selenium, vitamin D, essential fatty acids, and lipid profile

During illness:

- Call Metabolic Clinic to alert them immediately when you are sick
 - Poor feeding, fever, vomiting, or 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, phosphorous, ketones, CBC, amylase/lipase (risk of pancreatitis), possible ammonia, possible plasma amino acids, and possible urine organic acids
- IV carnitine may be given
- If possible, glycine should continue to be taken by mouth
- Please alert the Metabolic Clinic if you need surgery
- Wear Medic Alert Bracelet!!!**

Evaluations:

- Possible neurology evaluation if needed
- Possible brain MRI if needed

Topics:

- Review of biochemistry and genetics of isovaleric acidemia
- Diet/management
 - Leucine-restricted diet
 - Importance of medical formula
 - Introduction of low protein foods
 - Carnitine and glycine supplementation
 - Avoid fasting
 - Management during illness

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. If you urgently need to reach a Metabolic physician, call _____ and ask to page the Metabolic physician on call.

Thank You,
Your Metabolic Team