

Isovaleric Acidemia 1-18 years

Clinic Visits:

- At least every 6 months

Laboratory studies:

- Monitoring of diet
 - o plasma amino acid levels
 - 1-6 years: at least every 3 months
 - 6-18 years: at least every 6 months
- Yearly
 - o Comprehensive metabolic panel and phosphorous
 - o Amylase and lipase (risk of pancreatitis)
 - o Carnitine levels
 - o Prealbumin/albumin
 - o Ferritin, transferrin, or iron studies
 - o Possible urine organic acids
 - o Possible CBC, hemoglobin, and hematocrit
 - o Possible folate and vitamin B12
 - o Possible nutritional testing: zinc, selenium, vitamin D, essential fatty acids, and lipid profile

During illness:

- Call Metabolic Clinic to alert them immediately when your child is sick
 - o Poor feeding, fever, vomiting, 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: 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 to your child
- If possible, glycine should continue to be given by mouth
- 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 3 and 6 years old
- Neuropsychological evaluation at 9 years old
- Bone Health
 - o DEXA of spine at 9 and 18 years old
- Possible neurology evaluation if needed
- Possible brain MRI if needed

Topics:

- Review of biochemistry and genetics of isovaleric acidemia
- Diet/management
 - o Leucine-restricted diet

- Importance of medical formula
- Introduction of low protein foods
- Carnitine and glycine supplementation
- 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