

Glutaric Acidemia, Type I

Ages 2-18 years

Clinic Visits:

- 2- 12 years: at least every 6 months
- >12 years: at least yearly

Laboratory studies:

- Monitoring of diet
 - 2-6 years: plasma amino acid levels at least every 6 months
 - >6 years: plasma amino acid levels at least yearly
- At clinic visits:
 - Plasma amino acids
 - Possible glutaric and 3-hydroxyglutaric acid levels
 - Possible plasma acylcarnitine profile
- Yearly
 - Carnitine levels
 - Prealbumin/albumin
 - Ferritin, transferrin, or iron studies
 - Possible folate and vitamin B12 levels
 - Possible CBC, hemoglobin, and hematocrit
 - Possible nutritional testing: 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 and urine analysis for ketones
- IV carnitine may be given to your child
- 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
- Psychiatric screening at 18 years old
- DEXA scan (bone health) at 9 and 18 years old
 - If child does not bear weight, DEXA at 6, 9, 12, and 18 years
- Possible neurology evaluation if needed with possible head CT or brain MRI

Topics:

- Review of biochemistry and genetics of Glutaric Acidemia, Type I
- Diet/management
 - Lysine and tryptophan-restricted diet
 - Importance of medical formula

- Carnitine supplementation
- Possible riboflavin
- Avoid fasting
- Aggressive management during illness
- Medication for neurological symptoms if needed

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