

Glutaric Acidemia, Type I

Care Plan

| <p>Clinical Considerations</p> <ul style="list-style-type: none"> • Basal ganglia strokes • Retinal hemorrhages/ subdural hematomas (diff dx of NAT) • Macrocephaly • Low-excretors-may be missed on NBS | <p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> • UOA and ACP • Consider quant glutaric/3-OH glutaric • Consider molecular (sequencing) or Glutaryl-CoA dehydrogenase enzyme assay • Consider baseline head CT and/or brain MRI | | | | | | | | | | |
|--|---|-----------|------------|----------------|-------------|----------------|----------------|----------------|----------------|----------|--|
| <p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> • Dietary restriction of lysine and tryptophan • Formula/multivitamin • Carnitine- >100 mg/kg/day • Consider Riboflavin-100 mg/day • Neuropharmaceuticals if needed | <p>Monitoring</p> <ul style="list-style-type: none"> • Quant SAA <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;">Age</th> <th style="padding: 2px 5px;">Frequency</th> </tr> </thead> <tbody> <tr> <td style="padding: 2px 5px;">0-6 months</td> <td style="padding: 2px 5px;">Monthly</td> </tr> <tr> <td style="padding: 2px 5px;">6-24 months</td> <td style="padding: 2px 5px;">Every 3 months</td> </tr> <tr> <td style="padding: 2px 5px;">2-6 years</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> <tr> <td style="padding: 2px 5px;">>6 years</td> <td style="padding: 2px 5px;">Yearly</td> </tr> </tbody> </table> <p style="margin-left: 20px;">***Consider quant serum glutaric/3OH glutaric ***Consider ACP</p> | Age | Frequency | 0-6 months | Monthly | 6-24 months | Every 3 months | 2-6 years | Every 6 months | >6 years | Yearly |
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| <p>Frequency of visits</p> <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;">Age</th> <th style="padding: 2px 5px;">Frequency</th> </tr> </thead> <tbody> <tr> <td style="padding: 2px 5px;">0-6 months</td> <td style="padding: 2px 5px;">Every 2 months</td> </tr> <tr> <td style="padding: 2px 5px;">6-24 months</td> <td style="padding: 2px 5px;">Every 3 months</td> </tr> <tr> <td style="padding: 2px 5px;">2-12 years</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> <tr> <td style="padding: 2px 5px;">>12 years</td> <td style="padding: 2px 5px;">yearly</td> </tr> </tbody> </table> | Age | Frequency | 0-6 months | Every 2 months | 6-24 months | Every 3 months | 2-12 years | Every 6 months | >12 years | yearly | <p>Clinic visit labs</p> <ul style="list-style-type: none"> • See above • Quant SAA • Consider ACP |
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| 0-6 months | Every 2 months | | | | | | | | | | |
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| >12 years | yearly | | | | | | | | | | |
| <p>Emergency management</p> <ul style="list-style-type: none"> • Prevention of catabolism-stop protein • Immediate IV 10% dextrose plus lipids • IV carnitine, begin at 100 mg/kg/day • Aggressive fever management | <p>Labs to obtain during illness</p> <ul style="list-style-type: none"> • Basic metabolic panel • Urine ketones/UA | | | | | | | | | | |
| <p>Other evaluations</p> <ul style="list-style-type: none"> • Consider Baseline head CT and/or brain MRI • If MRI if not obtained at dx, then consider between 3-6 mo, with f/u MRI at 1, 2, 6, and 18 years if clinically indicated. • Nutrition eval with metabolic dietitian –freq? • Refer to Neurology if warranted • Bone health <ul style="list-style-type: none"> ○ DEXA-spine @ 9 & 18 y ○ Non weight bearing @ 6, 9, 12, and 18 y • Yearly developmental questionnaires (to be completed by parents) • Developmental eval @ 3 & 6 yrs • Neuropsych @ 9 y • Psychiatric screening at 18 years • Metabolic dietitian (at least yearly) | <p>Yearly labs</p> <ul style="list-style-type: none"> • Carnitine • Prealbumin / albumin • Plasma Ferritin, transferrin, or iron studies • Consider CBC, hemoglobin, and hematocrit • Folate and Vitamin B12 <ul style="list-style-type: none"> ○ If noncompliant with formula ○ Consider urine MMA • Consider other nutritional testing depending on formula (Zinc, selenium, vit D, essential fatty acids, and lipid profile). | | | | | | | | | | |



Glutaric Acidemia, Type I Care Plan

Performance Measures

1. Age of diagnosis (both positive NBD and confirmatory ACP and UOA)
2. Initial lab studies
 - a. NBS results
 - b. UOA and ACP
 - c. quant glutaric/3-OH glutaric
 - d. Molecular studies
 - e. Enzymatic studies
3. Monitoring lab studies
 - a. Quantitative serum amino acids (to monitor lysine)
 - b. Urine and/or serum glutaric/3-OH glutaric
 - c. Serum carnitine
 - d. Nutrition labs
4. Frequency of clinic contacts and visits (track compliance with visits)
5. Growth parameters (ht, wt, OFC, BMI)
6. Total decompensations and hospitalizations (including infections)
 - a. # of days for hospitalizations
 - b. # of ER visits
 - c. Track labs including BMP and urine ketones
7. Carnitine supplementation (Y/N, dosage)
8. Use of Neuropharmaceuticals for dystonia (Y/N, dosage and response)
9. DEXA results and number of fractures
10. MRI findings
11. Diet
 - a. Diet (Y/N)
 - b. Age of initiation
 - c. Age of discontinuation of diet (if applicable)
 - d. Frequency of dietitian visits (phone and clinic visits)
 - e. Frequency of dietary analysis (3 day diet records)
 - f. Natural protein intake (tolerance)
 - g. Lysine restriction or protein restriction
 - h. Formula (Y/N)
 - i. Medical foods (Y/N)
 - j. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)

12. Neuropsychological evaluation results
13. Developmental Services (PT, OT, & speech)
14. School Performance
 - a. Grade appropriate (Y/N)
 - b. IEP (Y/N)
 - c. Special services (Y/N)
14. Genetic Counseling (Y/N)

Outcome measures

1. Mortality
2. Development
 - a. IQ
 - b. Level of functioning
3. History and/or presence of neurological symptoms and abnormal MRI findings (basal ganglia changes)
4. History and/or presence of subdural hematomas
5. History and/or presence of retinal hemorrhages
6. Growth
 - a. Final adult parameters
 - b. Presence of macrocephaly