Mountain States Genetics
Regional Collaborative Center

Glutaric Acidemia, Type II (Neonatal/Infancy) Care Plan
(last updated 2/21/09)

Clinical Considerations

- Multiple acyl-CoA dehydrogenase deficiencies
- Three phenotypes
  - Neonatal with MCA-die within 1 week
  - Neonatal/Infantile w/o MCA-poor prognosis
  - Late or mild onset- usually normal uoa - see GA II mild care plan

Initial labs (diagnostic & baseline)

- UOA and ACP
- Carnitine
- CMP
- Ammonia
- Serum CK
- B-type Natriuretic Peptide (BNP)
- Enzymatic studies on cultured fibroblasts OR
- Sequence analysis of both genes

Diet considerations/ treatment

- Protein, fat, carbs modification
- Carnitine-begin at 100mg/kg/day
- Riboflavin-begin at 100mg/day
- Consider glycine
- Carb load when exercising
- Avoid fasting

Monitoring

- Plasma amino acids

Age	Frequency
0-6 months	Every 2 weeks
6-12 months	Monthly
1-6 years	Every 3 months
6-18 years	Every 6 months
>18 years	Yearly

Frequency of visits

Age	Frequency
0-6 months	Every 2 months
6-24 months	Every 3 months
>24 months	Every 6 months

Clinic visit labs

- Carnitine

Emergency management

- Immediate IV 10% dextrose
- IV carnitine, begin at 100 mg/kg/day

Labs to obtain during illness

- Comp metabolic panel
- Ammonia
- Serum CK
- BNP

Other evaluations

- Abdominal U/S at time of dx if clinically warranted
- Head CT at time of dx if clinically warranted
- Brain MRI yearly
- Echocardiogram yearly
- Yearly developmental questionnaires (to be completed by parents)
- Developmental eval @ 3 & 6y
- Neuropsych @ 9y
- Metabolic dietitian (at least yearly)

Yearly labs

- Comp metabolic panel
- Essential fatty acid profile @ 6 m then yearly thereafter
- BNP
- Prealbumin / albumin
- Plasma Ferritin, transferrin, or iron studies
- Consider CBC, hemoglobin, and hematocrit
- Folate and Vitamin B12
  - If noncompliant with formula
  - Consider urine MMA
- Consider other nutritional testing depending on formula (Zinc, selenium, vit D, and lipid profile).

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Performance Measures

1. Age of diagnosis and diet initiation
2. Presence of illness at time of diagnosis including hypotonia, tachypnea, metabolic acidosis, hepatomegaly, hypoglycemia, and sweaty feet odor.
3. Presence of congenital anomalies at time of diagnosis including hypotonia, hepatomegaly, hypoglycemia, metabolic acidosis, sweaty feet odor, kidneys are often palpably enlarged and cystic, facial dysmorphisms, rocker-bottom feet, muscular defects of the anterior abdominal wall and anomalies of the external genitalia (hypospadias and chordee).
4. Initial lab studies
    a. Acylcarnitine profile /UOA
    b. Other abnormal laboratory findings (if obtained) CMP, ammonia, CK, and BNP
    c. Carnitine
    d. Genotype
    e. Enzymatic studies
5. Frequency of clinic contacts and visits (track compliance with visits)
6. Growth parameters (ht, wt, OFC, BMI)
7. Total decompensations and hospitalizations (including infections)
    a. # of days for hospitalizations
    b. # of ER visits
    c. Track labs including CMP, ammonia, CK and BNP
8. Monitoring lab studies
    a. Plasma amino acids
    b. Carnitine
    c. Comp metabolic panel
    d. Nutrition labs
9. Carnitine supplementation (Y/N, dosage)
10. Riboflavin supplementation and responsiveness (Y/N, dosage)
11. Glycine supplementation and responsiveness
12. Use of 3-OH buterate
13. Diet
    a. Frequency of Dietitian visits
        i. Frequency of dietary analysis (3 day diet records)
    b. Natural protein intake (tolerance)
    c. Formula (Y/N)
    d. Medical foods (Y/N)
    e. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)
14. Neuropsychological evaluation results
15. Developmental services (PT, OT, & speech)
16. School Performance
    a. Grade appropriate (Y/N)
    b. IEP (Y/N)
    c. Special services (Y/N)
17. Genetic Counseling (Y/N)

Outcome measures

1. Mortality
2. History an/or presence of neuro symptoms
3. History and/or presence of liver and renal dysfunction
4. History and/or presence of cardiomyopathy
5. Development
    a. IQ
    b. Level of functioning
6. Growth
    a. Final adult parameters

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