Mountain States Genetics
Regional Collaborative Center

Glutaric Acidemia, Type II (Late or Mild onset) Care Plan
(last updated 2/21/09)

Clinical Considerations

- Multiple acyl-CoA dehydrogenase deficiencies –three phenotypes
- Late or mild onset- usually normal uoa
- Progressive lipid storage myopathy
- Carnitine deficiency
- Possible progressive extrapyramidal movement disorders

Initial labs (diagnostic & baseline)

- UOA and ACP
- Carnitine
- CMP
- Enzymatic studies on cultured fibroblasts OR
- Sequence analysis of both genes

Diet considerations/ treatment

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

Monitoring

- None if non-diet

Frequency of visits (if non diet)

Age	Frequency of clinic visits
0-4 years	Every 6 months
4-18 years	Once a year
>18 years	Every 3 years

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

Other evaluations

- Consider brain MRI if clinically warranted
- Yearly developmental questionnaires (to be completed by parents)
- Developmental eval @ 3 & 6y
- Neuropsych @ 9y

Yearly labs

- Comp metabolic panel

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Performance Measures

1. Age of diagnosis and diet initiation (if started)
2. Presence of illness at time of diagnosis including hypotonia, tachypnea, metabolic acidosis, hepatomegaly, hypoglycemia, and sweaty feet odor.
3. Initial lab studies
    a. Acylcarnitine profile /UOA
    b. Other abnormal laboratory findings (if obtained) CMP
    c. Carnitine
    d. Genotype
    e. Enzymatic studies
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 CMP
7. Monitoring lab studies
    a. Comp metabolic panel
    b. Carnitine levels
8. Carnitine supplementation (Y/N, dosage)
9. Riboflavin supplementation and responsiveness (Y/N, dosage)
10. Glycine supplementation and responsiveness (Y/N, dosage)
11. 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)
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)
15. Genetic Counseling (Y/N)

Outcome measures

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

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