Mountain States Genetics
Regional Collaborative Center

Carnitine-Acylcarnitine Translocase Deficiency Care Plan
(last updated 2/21/09)

Clinical Considerations

- Neonatal form with cardiomyopathy and arrhythmia (poor prognosis)
- Milder phenotype with hypoglycemia, but no cardiac
- Rare

Initial labs (diagnostic & baseline)

- ACP
- Consider UOA
- CACT sequencing (Horizon)
- R/O CPT II with sequencing
- Carnitine
- CMP
- Blood ammonia
- Serum CK

Diet considerations/ treatment

- Low-fat diet with limited long-chain triglycerides
- MCT oil
- High MCT formula
  - Need essential FA (safflower, walnut, canola)
- Consider carnitine
- Consider cornstarch (> 1 y) if symptomatic
- Avoid fasting
- If severe, continuous feeds at night

Monitoring

- Consider ACP

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

- Serum CK
- Carnitine panel
- LFTs
- Consider ACP

Emergency management

- Immediate IV 10% dextrose
- If oral carnitine not tolerated, switch to IV carnitine
- Consider cardiac monitoring (Chest X-ray to rule out cardiomyopathy, echo, EKG).

Labs to obtain during illness

- Comp metabolic panel
- Ammonia
- Serum CK
- BNP

Other evaluations

- Cardiology @ dx then every 2 years thereafter. Consider halter monitor
- R/O congenital anomalies at time of dx (due to CPT II in diff dx)
  - Abdominal U/S
  - Head CT
- Yearly developmental questionnaire (to be completed by parents)
- Developmental eval @ 3 & 6y
- Neuropsych @ 9y
- Metabolic dietitian (at least yearly)

Yearly labs

- Comprehensive metabolic panel
- ACP
- BNP
- Fatty acid profile at 6 m then yearly thereafter

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Performance Measures

1. Age of diagnosis and diet initiation
2. Presence of illness at time of diagnosis including lethargy, hypotonia, irritability, feeding difficulties, vomiting, hepatomegaly, tachypnea, cardiomyopathy, seizures, or coma.
3. Initial lab studies
    a. Acylcarnitine profile
    b. Other abnormal laboratory findings (if obtained) including CMP, ammonia, and CK
    c. Genotype
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, ammonia, CK, and BNP
7. Cardiac status at time of diagnosis and at follow-up
8. Monitoring laboratory studies
    a. ACP
    b. Comp metabolic panel
    c. Serum CK
    d. BNP
    e. Carnitine levels
    f. Fatty acid profile
9. Carnitine supplementation (Y/N, dosage)
10. Neuropsychological evaluation results
11. Diet
    a. Frequency of Dietitian visits
    b. Frequency of dietary analysis (3 day diet records)
    c. Percent of MCT and LCFA in diet.
        i. Including if essential fatty acids are added
    d. Formula (Y/N)
    e. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)
12. Developmental services (PT, OT, & speech)
13. 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. History and/or presence of cardiomyopathy and arrhythmia
3. History and/or presence of muscle fatigue and rhabdomyolysis
4. History and/or presence of myoglobinuria
5. History and/or presence of liver dysfunction
6. Development
    a. IQ
    b. Level of functioning
7. Growth
    a. Final adult parameters

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