Mountain States Genetics
Regional Collaborative Center

CPT II Deficiency Care Plan
(last updated 2/21/09)

Clinical Considerations

- Three distinct forms of CPT II deficiency:
- The common adult myopathic form
- The lethal neonatal form affecting many body systems (congenital anomalies)
- The severe infantile form which has liver, heart and skeletal involvement.

Initial labs (diagnostic & baseline)

- ACP
- Consider UOA
- Mutational/sequence analysis or enzymatic studies on fibroblasts
- Carnitine
- CMP
- Blood ammonia
- Serum CK
- BNP

Diet considerations/ treatment

- Low-fat diet with limited long-chain triglycerides for neonatal and infantile form
- MCT oil
- Portagen/Monogen
- High MCT formula
  - Need essential FA (safflower, walnut, canola)
- 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).
- Consider abdominal ultrasound

Labs to obtain during illness

- Comp metabolic panel
- Ammonia
- Serum CK
- Acylcarnitine profile
- BNP

Other evaluations

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

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 Laboratory Studies
    a. Acylcarnitine profile
    b. Other abnormal laboratory findings (if obtained) including CMP, ammonia, CK, and BNP
    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, ammonia, CK, ACP, and BNP
    d. Track rhabdomyolysis
7. Cardiac status at time of diagnosis and at follow-up
8. Monitoring lab studies
    a. ACP
    b. Comp metabolic panel
    c. Carnitine
    d. Serum CK
    e. BNP
9. Carnitine supplementation (Y/N, dosage)
10. 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)
11. Neuropsychological evaluation results
12. Developmental services (PT, OT, & speech)
13. 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 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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