

MCAD Deficiency Care Plan

(last updated 2/21/09)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> Risk underestimated in neonates 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> ACP Consider UOA Total/free carnitine Consider urine acylglycine Consider ACP/UOA on parents and sibs (if not previously screened) Consider mutational analysis should at least include the common mutation (A985G). <ul style="list-style-type: none"> If negative, proceed with sequence analysis Consider fatty acid ox flux studies (fibroblasts) 								
<p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> Avoid fasting Carnitine 50-100 mg/kg/day <ul style="list-style-type: none"> Increase dosage when sick Cornstarch if needed (>1 y when tolerate) Consider heart healthy diet (lower fat) Consider nutrition evaluation Carb load or increase carnitine prior to exercise Fasting tolerance Avoid excess MCT oil 	<p>Monitoring</p> <ul style="list-style-type: none"> Consider plasma carnitine levels at clinic visits 								
<p>Frequency of visits</p> <table border="1" data-bbox="154 1199 657 1339"> <thead> <tr> <th>Age</th> <th>Frequency of clinic visits</th> </tr> </thead> <tbody> <tr> <td>0-4 years</td> <td>Every 6 months</td> </tr> <tr> <td>4-18 years</td> <td>Once a year</td> </tr> <tr> <td>>18 years</td> <td>Every 3 years</td> </tr> </tbody> </table>	Age	Frequency of clinic visits	0-4 years	Every 6 months	4-18 years	Once a year	>18 years	Every 3 years	<p>Clinic visit labs</p> <ul style="list-style-type: none"> Consider carnitine
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<p>Emergency management</p> <ul style="list-style-type: none"> Immediate IV 10% dextrose Double oral carnitine dosage when sick If oral carnitine not tolerated, switch to IV carnitine Provide emergency letter Glucose gel and/or polycose 	<p>Labs to obtain during illness</p> <ul style="list-style-type: none"> Comp metabolic panel Consider ammonia Consider uric acid Consider urine analysis Consider serum CK 								
<p>Other evaluations</p> <ul style="list-style-type: none"> Yearly developmental questions (to be completed by parents) Developmental eval @ 3 & 6 yrs Neuropsych @ 9yrs 	<p>Yearly labs</p> <ul style="list-style-type: none"> None aside from carnitine at clinic visits 								

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Performance Measures	Outcome measures
<ol style="list-style-type: none"> 1. Age of diagnosis 2. Presence of illness at time of diagnosis including lethargy, vomiting, seizures, and coma 3. Initial lab studies <ol style="list-style-type: none"> a. Acylcarnitine profile b. Other abnormal laboratory findings (if obtained) including glucose, ammonia, uric acid, and LFTS c. Carnitine d. Acylglycines e. Genotype f. Enzymatic studies 4. Frequency of clinic contacts and visits (track compliance with visits) 5. Growth parameters (ht, wt, OFC, BMI) 6. Monitoring labs <ol style="list-style-type: none"> a. Carnitine 7. Total decompensations and hospitalizations (including infections) <ol style="list-style-type: none"> a. # of days for hospitalizations b. # of ER visits c. Track labs including CMP, ammonia, uric acid, urine ketones, and CK 8. Carnitine supplementation (Y/N, dosage) 9. Use of cornstarch (Y/N, dosage) 10. Exercise intolerance/fatigue (Y/N) 11. Sports participation (type, level) 12. Diet <ol style="list-style-type: none"> a. Frequency of Dietitian visits b. Frequency of dietary analysis (3 day diet records) c. Modified diet (Y/N) d. Percentage of fat in diet 13. Neuropsychological evaluation results 14. Developmental services (PT, OT, & speech) 15. School performance <ol style="list-style-type: none"> a. Grade appropriate (Y/N) b. IEP (Y/N) c. Special services (Y/N) 16. Genetic Counseling (Y/N) 	<ol style="list-style-type: none"> 1. Mortality 2. History and/or presence of obesity and related secondary complications (insulin resistance and type II diabetes) 3. History and/or presence of fatigue and muscle pain 4. History and/or presence of reduced exercise intolerance /fatigue 5. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 6. Growth <ol style="list-style-type: none"> a. Final adult parameters