

LCHAD and TFP Deficiencies Care Plan

(last updated 2/21/09)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> • Cardiomyopathy and arrhythmia • Carrier women of affected fetus can have HELLP syndrome and acute fatty liver of pregnancy • Pigmentary retinopathy • Risk for neuropathy • Risk for rhabdomyolysis and myoglobinuria • Risk for hepatic dysfunction 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> • ACP • Consider UOA • Consider Mutational analysis • Consider FAO flux studies • Carnitine • Comp metabolic panel • Serum CK • Consider B-type Natriuretic Peptide (BNP) 								
<p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> • Low-fat diet with limited long-chain triglycerides • MCT oil or high MCT formula <ul style="list-style-type: none"> ◦ Need essential FA (safflower, walnut, canola) • Consider Carnitine • Consider Cornstarch (> 1 y) if symptomatic • Avoid fasting • Exercise with physician consultation. Consider carb load. 	<p>Monitoring</p> <ul style="list-style-type: none"> • See clinic visits 								
<p>Frequency of visits</p> <table border="1" data-bbox="170 955 581 1102"> <thead> <tr> <th>Age</th> <th>Frequency</th> </tr> </thead> <tbody> <tr> <td>0-6 months</td> <td>Every 2 months</td> </tr> <tr> <td>6-24 months</td> <td>Every 3 months</td> </tr> <tr> <td>>24 months</td> <td>Every 6 months</td> </tr> </tbody> </table>	Age	Frequency	0-6 months	Every 2 months	6-24 months	Every 3 months	>24 months	Every 6 months	<p>Clinic visit labs</p> <ul style="list-style-type: none"> • Consider carnitine • Consider serum CK • Consider ACP • Consider LFTs
Age	Frequency								
0-6 months	Every 2 months								
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<p>Emergency management</p> <ul style="list-style-type: none"> • Immediate IV D5-D10% dextrose plus IV fluids (hyper-hydration) • If on oral carnitine & not tolerated, switch to IV carnitine • If rhabdomyolysis, hyperhydrate +/- alkalinization • Consider cardiac monitoring (Chest X-ray to rule out cardiomyopathy, echo, EKG). 	<p>Labs to obtain during illness</p> <ul style="list-style-type: none"> • Comp metabolic panel • Consider ammonia • Serum CK • Acylcarnitine profile • BNP 								
<p>Other evaluations</p> <ul style="list-style-type: none"> • Cardiac eval @ dx , 6 mo, 12 mo, yearly • Ophthalmology yearly (retinal exam) • Nerve conduction/velocity studies if clinically warranted. • Referral to neurology if peripheral neuropathy noted on PE • Yearly developmental questionnaires (to be completed by parents) • Developmental eval @ 3 & 6y • Neuropsych @ 9y • Psychiatric screening @ 18y • Metabolic dietitian (at least yearly) 	<p>Yearly labs</p> <ul style="list-style-type: none"> • Comprehensive metabolic panel • ACP • BNP • Essential fatty acid profile @ 6 m then yearly thereafter • Consider nutritional labs 								

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<p>Performance Measures</p> <ol style="list-style-type: none"> 1. Age of diagnosis and diet initiation 2. Presence of maternal complications during pregnancy (HELLP, AFLP, preeclampsia) 3. Presence of illness at time of diagnosis including lethargy, hypotonia, irritability, feeding difficulties, vomiting, hepatomegaly, tachypnea, cardiomyopathy, seizures, or coma. 4. Initial lab studies <ol style="list-style-type: none"> a. Acylcarnitine profile b. Other abnormal laboratory findings (if obtained) including glucose, electrolytes, ammonia, and LFTs. 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) <ol style="list-style-type: none"> a. # of days for hospitalizations b. # of ER visits c. Track labs including ACP, CMP, ammonia, CK, and BNP 8. Cardiac status at time of diagnosis and thereafter 9. Monitoring lab studies <ol style="list-style-type: none"> a. LFTs b. Kidney function c. Carnitine d. Fatty acid profile e. BNP 10. Carnitine supplementation (Y/N,dosage) 11. Diet <ol style="list-style-type: none"> a. Frequency of Dietitian visits b. Frequency of dietary analysis (3 day diet records) c. Percent of MCT and LCFA in diet. <ol style="list-style-type: none"> i. Including if essential fatty acids are added d. Formula (Y/N) e. Mode (oral, G-tube, bolus/drip, meds only/meds and diet) 	<ol style="list-style-type: none"> 12. Ophthalmology evals 13. Exercise intolerance/fatigue (Y/N) 14. Sports participation (type and level) 15. Neuropsychological evaluation results 16. Developmental services (PT, OT, & speech) 17. School performance <ol style="list-style-type: none"> a. Grade appropriate (Y/N) b. IEP (Y/N) c. Special services (Y/N) 18. Genetic Counseling (Y/N) <p>Outcome measures</p> <ol style="list-style-type: none"> 1. Mortality 2. History of hypoglycemic episodes 3. History and/or presence of cardiomyopathy 4. History and/or presence of muscle fatigue, rhabdomyolysis, and exercise intolerance 5. History and/or presence of myoglobinuria 6. History and/or presence of liver dysfunction 7. History and/or presence of pigmentary retinopathy 8. History and/or presence of neuropathy. 9. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 10. Growth <ol style="list-style-type: none"> a. Final adult parameters
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