

VLCAD Deficiency 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 • Hepatic dysfunction • Risk for rhabdomyolysis and myoglobinuria • Hypoglycemia • Normalization of ACP following +NBS in affected | <p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> • ACP • Consider UOA • Sequence analysis • Consider FAO flux studies if no or only one mutation found • Comp metabolic panel • Carnitine • Serum CK • UA • B-type Natriuretic Peptide (BNP) | | | | | | | | |
|--|--|------------------|------------|----------------|-------------|----------------|------------|----------------|--|
| <p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> • Low-fat diet with limited long-chain triglycerides and supplemented MCT oil • Can be liberalized based on genotype and ACP • 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"> • Consider ACP | | | | | | | | |
| <p>Frequency of visits</p> <table border="1" style="margin-left: auto; margin-right: auto;"> <thead> <tr> <th style="text-align: center;"><u>Age</u></th> <th style="text-align: center;"><u>Frequency</u></th> </tr> </thead> <tbody> <tr> <td style="text-align: center;">0-6 months</td> <td style="text-align: center;">Every 2 months</td> </tr> <tr> <td style="text-align: center;">6-24 months</td> <td style="text-align: center;">Every 3 months</td> </tr> <tr> <td style="text-align: center;">>24 months</td> <td style="text-align: center;">Every 6 months</td> </tr> </tbody> </table> | <u>Age</u> | <u>Frequency</u> | 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"> • Carnitine • Serum CK • Consider ACP • Consider LFTs |
| <u>Age</u> | <u>Frequency</u> | | | | | | | | |
| 0-6 months | Every 2 months | | | | | | | | |
| 6-24 months | Every 3 months | | | | | | | | |
| >24 months | Every 6 months | | | | | | | | |
| <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, use myoglobinuria protocol • 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 • Ammonia • Serum CK • Acylcarnitine profile • BNP | | | | | | | | |
| <p>Other evaluations</p> <ul style="list-style-type: none"> • Echocardiogram @ dx , 6 mo, 12 mo, yearly thereafter • Yearly developmental questionnaires (to be completed by parents) • Developmental eval @ 3 & 6y • Neuropsych @ 9y • 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 | | | | | | | | |

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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. Normalization of values (Y/N) c. Other abnormal laboratory findings (if obtained) including CMP, CK, UA, and BNP 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 CMP, ammonia, ACP, CK, and BNP d. Track rhabdomyolysis 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) 12. Neuropsychological evaluation results 13. Developmental services (PT, OT, & speech) | <ol style="list-style-type: none"> 14. School Performance <ol style="list-style-type: none"> a. Grade appropriate (Y/N) b. IEP (Y/N) c. Special services (Y/N) 15. 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 <ol style="list-style-type: none"> 1. History and/or presence of muscle fatigue and rhabdomyolysis 2. History and/or presence of myoglobinuria 3. History and/or presence of liver dysfunction 4. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 5. Growth <ol style="list-style-type: none"> a. Final adult parameters |
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