

Beta-ketothiolase deficiency Care Plan (updated 7/13/10)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> • Metabolic acidosis • Clinical heterogeneity • Neuro involvement as sequelae of ketoacidosis • Neutropenia and thrombocytopenia • Poor weight gain, renal failure, and short stature can occur 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> • UOA and ACP • CMP • CBC • Acetoacetyl –CoA thiolase enzyme assay and/or molecular analysis of ACAT1 gene 																
<p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> • Mild protein restriction (1.5-2.0 g/kg/day) • Carnitine- 50-100 mg/kg/day • Avoidance of fasting • Consider cornstarch (> 6 m) 	<p>Monitoring</p> <ul style="list-style-type: none"> • Pl AA for diet patient <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;">Age</th> <th style="padding: 2px 5px;">Frequency</th> </tr> </thead> <tbody> <tr> <td style="padding: 2px 5px;">0-6 months</td> <td style="padding: 2px 5px;">Every 2 weeks</td> </tr> <tr> <td style="padding: 2px 5px;">6-12 months</td> <td style="padding: 2px 5px;">Monthly</td> </tr> <tr> <td style="padding: 2px 5px;">1-6 years</td> <td style="padding: 2px 5px;">Every 3 months</td> </tr> <tr> <td style="padding: 2px 5px;">6-18 years</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> <tr> <td style="padding: 2px 5px;">>18 years</td> <td style="padding: 2px 5px;">Yearly</td> </tr> </tbody> </table>	Age	Frequency	0-6 months	Every 2 weeks	6-12 months	Monthly	1-6 years	Every 3 months	6-18 years	Every 6 months	>18 years	Yearly				
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<p>Frequency of visits</p> <ul style="list-style-type: none"> • no diet <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;">Age</th> <th style="padding: 2px 5px;">Frequency</th> </tr> </thead> <tbody> <tr> <td style="padding: 2px 5px;">0-4 years</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> <tr> <td style="padding: 2px 5px;">4-18 years</td> <td style="padding: 2px 5px;">Once a year</td> </tr> <tr> <td style="padding: 2px 5px;">>18 years</td> <td style="padding: 2px 5px;">Every 3 years</td> </tr> </tbody> </table> <ul style="list-style-type: none"> • Diet <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;">Age</th> <th style="padding: 2px 5px;">Frequency</th> </tr> </thead> <tbody> <tr> <td style="padding: 2px 5px;">0-6 months</td> <td style="padding: 2px 5px;">Every 2 months</td> </tr> <tr> <td style="padding: 2px 5px;">6-12 months</td> <td style="padding: 2px 5px;">Every 3 months</td> </tr> <tr> <td style="padding: 2px 5px;">>12 months</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> </tbody> </table>	Age	Frequency	0-4 years	Every 6 months	4-18 years	Once a year	>18 years	Every 3 years	Age	Frequency	0-6 months	Every 2 months	6-12 months	Every 3 months	>12 months	Every 6 months	<p>Clinic visit labs</p> <ul style="list-style-type: none"> • Carnitine • Consider UOA • Consider CMP
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<p>Emergency management</p> <ul style="list-style-type: none"> • Immediate IV 10% dextrose + bicarb • IV carnitine • Consider IV lipids if fasting > 12 hours 	<p>Labs to obtain during illness</p> <ul style="list-style-type: none"> • CMP • Ammonia • Urine ketones • Renal studies (BUN, creatinine, calcium, phosphorous) 																
<p>Other evaluations</p> <ul style="list-style-type: none"> • If neuro symptoms present, referral to neurology with brain MRI warranted • Yearly developmental questionnaires (to be completed by parents). • Developmental eval @ 3 & 6y • Neuropsych @ 9y • For diet patients, metabolic dietitian (at least yearly) 	<p>Yearly labs</p> <ul style="list-style-type: none"> • CMP and phosphate • CBC • Serum carnitine • Renal studies (BUN, creatinine, calcium, phosphorous) <p>If on diet:</p> <ul style="list-style-type: none"> • Prealbumin / albumin • Plasma Ferritin, transferrin, or iron studies • Consider folate and Vitamin B12 (if noncompliant with formula) • Consider other nutritional testing depending on formula (Zinc, selenium, vit D, essential fatty acids, and lipid profile). 																

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<p>Performance Measures</p> <ol style="list-style-type: none"> 1. Age of diagnosis (both positive NBS and confirmatory ACP and UOA) 2. Presence of illness at time of diagnosis (neonatal presentation rare) 3. Initial lab studies <ol style="list-style-type: none"> a. NBS results b. Acylcarnitine profile c. Urine organic acids d. Quant plasma amino acids e. Metabolic panel f. Ammonia g. LFTs h. Molecular studies i. Enzymatic studies 4. Monitoring lab studies <ol style="list-style-type: none"> a. Quant plasma amino acids b. Urine organic acids c. Carnitine d. Metabolic panel e. Nutrition labs 5. Frequency of clinic contacts and visits (track compliance with visits) 6. Growth parameters (ht, wt, OFC, BMI) 7. Total decompensations and hospitalizations <ol style="list-style-type: none"> a. # of days for hospitalizations b. # of ER visits c. Track labs including CMP, ammonia ketones, and renal studies 8. EKG and cardiac findings 9. Carnitine supplementation (Y/N, dosage) 10. Diet (Y/N) <ol style="list-style-type: none"> a. Frequency of Metabolic dietitian visits b. Frequency of dietary analysis (3 day diet records) c. Natural protein intake (tolerance) d. Formula (Y/N) e. Medical foods (Y/N) f. Mode 	<ol style="list-style-type: none"> 11. Neuropsychological evaluation results 12. Developmental services (PT, OT, & speech) 13. School Performance <ol style="list-style-type: none"> a. Grade appropriate (Y/N) b. IEP (Y/N) c. Special services (Y/N) 14. Genetic Counseling (Y/N) <p>Outcome measures</p> <ol style="list-style-type: none"> 1. Mortality 2. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 3. History and/or presence of cardiomyopathy and Long QT syndrome 4. History and/or presence of neurological symptoms and abnormal MRI findings (basal ganglia changes) 5. History and/or presence of renal dysfunction/failure. 6. Growth <ol style="list-style-type: none"> a. Final adult parameters
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