

Methylmalonic Acidemia –Mutase, Cbl A & Cbl B Care Plan (last updated 2/21/09)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> • Stabilize neonate • Pancreatitis • Bone marrow suppression • Renal disease • B12 responsive • Cardiomyopathy 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> • UOA and SAA • ACP • Comp metabolic panel, ammonia, CBC, urine ketones, tHcy, B12 • Consider quant MMA • Complementation studies and/or molecular analysis 												
<p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> • Dietary restriction of propiogenic amino acids • Essential amino acid formula • B12- 1 mg/day for 2 week trial (IM or subcut) • Carnitine- 50-100 mg/kg/day • Antibiotics for gut flora if poor control • Consider liver and/or kidney transplant • Avoidance of fasting 	<p>Monitoring</p> <ul style="list-style-type: none"> • Quant SAA and Quant MMA <table border="1" style="margin-left: 20px; border-collapse: collapse; text-align: center;"> <thead> <tr> <th style="padding: 2px 5px;"><u>Age</u></th> <th style="padding: 2px 5px;"><u>Frequency</u></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>	<u>Age</u>	<u>Frequency</u>	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>Emergency management</p> <ul style="list-style-type: none"> • Immediate IV 10% dextrose • Consider IV lipids if fasting >12 hours • Add IV carnitine 100 mg/kg/day • Consider cardiac monitoring 	<p>Labs to obtain during illness</p> <ul style="list-style-type: none"> • Comp metabolic panel and phosphate • CBC • Urinalysis for ketones • Amylase and lipase • Consider ammonia • Consider SAA and quant MMA • Consider B-type Natriuretic peptide- BNP 												
<p>Other evaluations</p> <ul style="list-style-type: none"> • Referral to neurology if clinically warranted. Consider brain MRI. • Bone health <ul style="list-style-type: none"> ○ DEXA –spine @ 9 & 18 y • Yearly developmental questionnaires (to be completed by parents). • Developmental eval @ 3 & 6 y • Neuropsych @ 9 y • Consider renal eval • Consider cardiology eval (especially in illness) • Metabolic dietitian (at least yearly) 	<p>Yearly labs</p> <ul style="list-style-type: none"> • Comp metabolic panel • Urinalysis for protein • Blood pressure • Serum carnitine • SAA, UOA, and Quant urine/plasma MMA • Amylase and lipase • Prealbumin / albumin • Plasma Ferritin, transferrin, or iron studies • Consider 24 hour urine creatinine clearance and protein @ 6 y then yearly thereafter • Consider BNP • Consider CBC, hemoglobin, and hematocrit • 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 including poor feeding, vomiting, severe metabolic acidosis, ketosis, ketonuria, hyperammonemia, and hyperglycemia. 3. Initial lab studies <ol style="list-style-type: none"> a. NBS results b. UOA c. SAA d. ACP e. CMP f. Ammonia g. CBC h. urine ketones i. tHcy j. B12 k. Quant MMA l. Complementation studies m. Molecular analysis 4. Monitoring lab studies <ol style="list-style-type: none"> a. Quantitative branched-chain amino acids . b. Metabolic panel c. Serum carnitine d. Renal studies e. Nutrition labs 5. Total decompensations and hospitalizations <ol style="list-style-type: none"> a. # of days for hospitalizations b. # of ER visits c. Track ICU admissions d. Track labs including CMP, phosphate, ketones, amylase/lipase, ammonia, SAA, MMA, and BNP 6. Frequency of clinic contacts and visits (track compliance with visits) 7. B12 responsiveness (Y/N, dosage) 8. Growth parameters (ht, wt, OFC, BMI) 9. Number of pancreatitis episodes 10. Dosage of carnitine supplementation 11. Use of antibiotics (Y,N, type, and dosage) 12. DEXA results and number of bone fractures 13. Transplant <ol style="list-style-type: none"> a. Liver (Y/N) b. Kidney (Y/N) 	<ol style="list-style-type: none"> 14. Diet <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 (oral, G-tube, bolus/drip, meds only/meds and diet) 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. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 3. History and/or presence of neurological symptoms (dystonia) and abnormal MRI findings (basal ganglia changes) 4. History and/or presence of renal dysfunction and failure. 5. History and/or presence of cardiac complications 6. History and/or presence of pancreatitis 7. History and /or presence of osteopenia. 8. Outcome of renal and/or liver transplant 9. Growth <ol style="list-style-type: none"> a. Final adult parameters
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