

Cbl C, Cbl D, & Cbl F (MMA with HCY) Care Plan (last updated 7/13/10)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> Hematologic manifestations (pancytopenia, macrocytic or microcytic anemia) Renal and hepatic dysfunction Neurological involvement Vision impairment Cardiac involvement Feeding difficulties, FTT, hyponatremia, developmental delays 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> UOA and pIAA ACP Comp metabolic panel, CBC, tHcy, B12 Consider quant MMA Complementation studies and/or molecular analysis 												
<p>Diet considerations/ treatment</p> <ul style="list-style-type: none"> Hydroxocobalamin IM/IC Carnitine - 50-100 mg/kg/day Betaine – 100-250 mg/kg/day Folinic acid- 25 mg Consider Matanx (methylcbl, methylfolate, and pyridoxine) – 2 tab/day Consider diet 	<p>Monitoring</p> <ul style="list-style-type: none"> Serum or urine MMA, pl total Hcy, qt Met Met >25 umol/L and tHcy < 50 umol/L <table border="1" style="margin-left: 40px; 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 bicarb Consider IV or IM hydroxocobalamin 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 Urine ketones quant MMA total homocysteine Consider B-type Natriuretic peptide- BNP Consider carnitine, pl AA and UOA 												
<p>Other evaluations</p> <ul style="list-style-type: none"> Ophthalmology eval at time of dx and yearly thereafter. Consider electroretinogram. Consider Cardiology if clinically warranted Referral to neurology if clinically warranted. Consider brain MRI. Consider renal eval if clinically warranted. Consider liver eval if clinically warranted If on diet, DEXA -spine @ 9 & 18 years If on diet, metabolic dietitian yearly Yearly developmental questionnaires (to be completed by parents). Developmental eval @ 3 & 6 y Neuropsych @ 9 y 	<p>Yearly labs</p> <ul style="list-style-type: none"> Comp metabolic panel Urinalysis for protein 24 hour urine creatinine clearance and protein (calculated GFR) Blood pressure CBC, hemoglobin, and hematocrit Serum carnitine Consider BNP <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 1. Presence of illness at time of diagnosis including poor feeding, hypotonia, metabolic acidosis, cardiomyopathy, hematological findings, and renal involvement. 2. Initial lab studies <ol style="list-style-type: none"> a. NBS results b. UOA c. ACP d. CMP e. CBC f. tHcy g. B12 h. Quant MMA i. Complementation studies j. Molecular analysis 3. Monitoring lab studies <ol style="list-style-type: none"> a. Serum methylmalonic acid b. Plasma total homocysteine c. Renal studies 4. 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 5. Frequency of clinic contacts and visits (track compliance with visits) 6. Hydroxocobalamin (dosage) 7. Betaine (dosage) 8. Growth parameters (ht, wt, OFC, BMI) 9. Dosage of carnitine supplementation 10. 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) 11. DEXA results and number of bone fractures 	<ol style="list-style-type: none"> 12. Neuropsychological evaluation results 13. Developmental services (PT, OT, & speech) 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. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning 3. History and/or presence of hematologic manifestations (pancytopenia, macrocytic or microcytic anemia, hypersegmented neutrophils, and thrombocytopenia) 4. History and/or presence of ophthalmologic problems (retinopathy, abnl ocular movements, visual acuity) 5. History and/or presence of neurological symptoms (seizures, spasticity, microcephaly, hydrocephalus, abnormal gait) and abnormal MRI findings (white matter loss, basal ganglia changes) 6. History and/or presence of renal dysfunction and failure. 7. History and/or presence of hepatic dysfunction 8. History and/or presence of hemolytic uremic syndrome 9. History and/or presence of cardiac complications (congestive heart failure, cardiomyopathy) 10. Growth 11. Final adult parameters
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