

Hypermethioninemia (MAT -1) Care Plan

(last updated 2/21/09)

<p>Clinical Considerations</p> <ul style="list-style-type: none"> Majority are asymptomatic Diagnosis of exclusion 	<p>Initial labs (diagnostic & baseline)</p> <ul style="list-style-type: none"> Quant plasma amino acids Total homocysteine Liver function and PT/PTT Consider sequence analysis (available on research basis) 																		
<p>Diet Considerations</p> <ul style="list-style-type: none"> S-adenosyl-L-methionine (AdoMet/SAM-e) supplementation if brain MRI is abnormal. Met restriction (targeted met level <1000μmol/L) 	<p>Monitoring labs</p> <ul style="list-style-type: none"> See clinic visit labs 																		
<p>Frequency of metabolic visits</p> <p>Non-diet</p> <table border="1" data-bbox="155 781 537 919"> <thead> <tr> <th>Age</th> <th>Frequency</th> </tr> </thead> <tbody> <tr> <td>0-4 years</td> <td>Every 6 months</td> </tr> <tr> <td>4-18 years</td> <td>Once a year</td> </tr> <tr> <td>>18 years</td> <td>Every 3 years</td> </tr> </tbody> </table> <p>Diet</p> <table border="1" data-bbox="155 951 545 1121"> <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>2-18 years</td> <td>Every 6 months</td> </tr> <tr> <td>>18 years</td> <td>Yearly</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	6-24 months	Every 3 months	2-18 years	Every 6 months	>18 years	Yearly	<p>Clinic Visit labs</p> <ul style="list-style-type: none"> Quant plasma amino acids <ul style="list-style-type: none"> Targeted met <1000 Consider Total homocysteine S-adenosyl-L-methionine (AdoMet/SAM-e) levels if possible Liver function
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<p>Other evaluations</p> <ul style="list-style-type: none"> Baseline MRI @ 1 year then F/U brain MRI every 3 yrs If abnormal MRI or child is symptomatic, consider yearly MRIs Development eval @ 3 and 6y Neuropsych @ 9 y For diet patients, metabolic dietitian eval (at least yearly) 	<p>Yearly labs</p> <p>***If on Diet:</p> <ul style="list-style-type: none"> Prealbumin / albumin Plasma Ferritin, transferrin, or iron studies Consider CBC, hemoglobin, and hematocrit Consider folate and vitamin B12 Consider other nutritional testing (Zinc, selenium, vit D, essential fatty acids, and lipid profile). 																		

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Performance Indicators	Outcome Indicators
<ol style="list-style-type: none"> 1. Frequency of clinic contacts and visits (track compliance with visits) 2. Initial lab studies <ol style="list-style-type: none"> a. NBS results b. Quant plasma amino acids c. Plasma total homocysteine d. LFTs e. PT/PTT f. SAM and SAH levels g. Molecular studies 3. Monitoring lab studies <ol style="list-style-type: none"> a. Quant plasma amino acids b. Plasma total homocysteine c. LFTs d. SAM e. Nutrition labs (if on diet) 4. Age of initiation of SAME supplementation (track pre/post serum levels, dosage) 5. Growth parameters (ht, wt, OFC, BMI) 6. MRI findings 7. Neuropsychological evaluation 8. If on 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 9. School performance <ol style="list-style-type: none"> a. Grade appropriate (Y/N) b. Special services (Y/N) c. IEP (Y/N) 10. Genetic Counseling (Y/N) 	<ol style="list-style-type: none"> 1. History and/or presence of neurological disease 2. History and/or presence of demyelination 3. History and/or presence of liver dysfunction 4. Development <ol style="list-style-type: none"> a. IQ b. Level of functioning c. Decline in IQ or level of function 5. Growth <ol style="list-style-type: none"> a. Final adult parameters