

## Homocystinuria Care Plan

(last updated 2/21/09)

<p><b>Clinical Considerations</b></p> <ul style="list-style-type: none"> <li>• Virtually all infants detected by NBS are pyridoxine non-responsive</li> <li>• Pyridoxine responsiveness</li> <li>• Thrombosis</li> <li>• Psychiatric problems</li> <li>• Eye-dislocated lens</li> </ul>	<p><b>Initial labs (diagnostic &amp; baseline)</b></p> <ul style="list-style-type: none"> <li>• Quant plasma AA, UOA, and total plasma homocysteine</li> <li>• Consider cystathionine <math>\beta</math>-synthase (CBS) enzyme activity (cultured fibroblasts)</li> <li>• Consider molecular-majority are private, sequencing available</li> <li>• Consider MTHFR and factor V testing</li> </ul>												
<p><b>Diet considerations/ treatment</b></p> <ul style="list-style-type: none"> <li>• Met restricted diet w/ Met-free formula</li> <li>• L-cysteine supplementation if low</li> <li>• Folate 1 mg/day <ul style="list-style-type: none"> <li>○ If MTHFR carrier, 5-10 mg/day</li> </ul> </li> <li>• B12 oral 1mg/day</li> <li>• Betaine</li> <li>• Aspirin if &gt;12 yrs</li> <li>• Prophylactic anticoagulation during pregnancy</li> </ul>	<p><b>Monitoring</b></p> <ul style="list-style-type: none"> <li>• Quant pl amino acids and total homocysteine</li> <li>• Targeted levels (<math>\mu</math>mol/L) <ul style="list-style-type: none"> <li>○ Homocysteine &lt;50</li> <li>○ Methionine &lt;1000</li> </ul> </li> </ul> <table border="1" data-bbox="917 703 1377 913"> <thead> <tr> <th>Age</th> <th>Frequency</th> </tr> </thead> <tbody> <tr> <td>0-6 months</td> <td>Every 2 weeks</td> </tr> <tr> <td>6-12 months</td> <td>Monthly</td> </tr> <tr> <td>1-6 years</td> <td>Every 3 months</td> </tr> <tr> <td>6-18 years</td> <td>Every 6 months</td> </tr> <tr> <td>&gt;18 years</td> <td>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><b>Frequency of visits</b></p> <table border="1" data-bbox="224 961 730 1102"> <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 m-18 years</td> <td>Every 6 months</td> </tr> <tr> <td>&gt;18 years</td> <td>Yearly</td> </tr> </tbody> </table> <p>***same for B6 responsive patients</p>	Age	Frequency	0-6 months	Every 2 months	6 m-18 years	Every 6 months	>18 years	Yearly	<p><b>Clinic visit labs</b></p> <ul style="list-style-type: none"> <li>• Quant plasma amino acids</li> <li>• Total homocysteine</li> </ul>				
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<p><b>Emergency management</b></p> <ul style="list-style-type: none"> <li>• ER letter for thrombosis risks with surgery and illness</li> </ul>	<p><b>Labs to obtain during illness</b></p> <ul style="list-style-type: none"> <li>• None</li> </ul>												
<p><b>other evaluations</b></p> <ul style="list-style-type: none"> <li>• Ophthalmology-yearly</li> <li>• Neurology eval if clinically warranted</li> <li>• Bone health <ul style="list-style-type: none"> <li>○ DEXA-spine 6, 9, 12, &amp; 18 y (then every 5 years)</li> </ul> </li> <li>• Yearly developmental questionnaires (to be completed by parents)</li> <li>• Developmental eval @ 3 &amp; 6 y</li> <li>• Neuropsych @ 9y</li> <li>• Psychiatric screening</li> <li>• Refer to Orthopedics if clinically warranted</li> <li>• Metabolic dietitian eval (at least yearly)</li> </ul>	<p><b>Yearly labs</b></p> <ul style="list-style-type: none"> <li>• Prealbumin / albumin</li> <li>• Plasma Ferritin, transferrin, or iron studies</li> <li>• Consider CBC, hemoglobin, and hematocrit</li> <li>• Consider folate and vitamin B12</li> <li>• Consider other nutritional testing (Zinc, selenium, vit D, essential fatty acids, and lipid profile).</li> </ul>												

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<p><b>Performance Measures</b></p> <ol style="list-style-type: none"> <li>1. Age of initiation of diet</li> <li>2. B6 responsiveness and dosage at which responsive</li> <li>3. Initial lab studies             <ol style="list-style-type: none"> <li>a. NBS results</li> <li>b. Quant plasma amino acids</li> <li>c. Urine organic acids</li> <li>d. Plasma total homocysteine</li> <li>e. Molecular studies</li> <li>f. Enzymatic studies</li> <li>g. MTHFR and factor V (if performed)</li> </ol> </li> <li>4. Monitoring lab studies             <ol style="list-style-type: none"> <li>a. Quant plasma amino acids</li> <li>b. Plasma total homocysteine</li> <li>c. Nutrition labs</li> </ol> </li> <li>5. Frequency of clinic contacts and visits (track compliance with visits)</li> <li>6. Betaine treatment (track dosage)</li> <li>7. Growth parameters (ht, wt, OFC, BMI)</li> <li>8. Ophthalmology evaluations</li> <li>9. Track all vascular incidents (type, age, homocysteine levels, and medications at time of presentation).</li> <li>10. Number of pancreatitis episodes</li> <li>11. Neurology evaluation for cerebral edema or neuropathy</li> <li>12. DEXA results and number of fractures</li> <li>13. Diet             <ol style="list-style-type: none"> <li>a. Frequency of Metabolic dietitian visits</li> <li>b. Frequency of dietary analysis (3 day diet records)</li> <li>c. Natural protein intake (tolerance)</li> <li>d. Formula (Y/N)</li> <li>e. Medical foods (Y/N)</li> <li>f. Mode</li> </ol> </li> </ol>	<ol style="list-style-type: none"> <li>14. Neuropsychological evaluation</li> <li>15. Psychiatric screening</li> <li>16. Developmental services (PT, OT, &amp; speech)</li> <li>17. School performance             <ol style="list-style-type: none"> <li>a. Grade appropriate (Y/N)</li> <li>b. Special services (Y/N)</li> <li>c. IEP (Y/N)</li> </ol> </li> <li>18. Genetic Counseling (Y/N)</li> </ol> <p><b>Outcome measures</b></p> <ol style="list-style-type: none"> <li>1. History and/or presence of ectopia lentis and myopia</li> <li>2. History of vascular incidents</li> <li>3. History and/or presence of neurological complications (seizures, cerebral edema, and peripheral neuropathy)</li> <li>4. History and/or presence of pancreatitis</li> <li>5. History and/or presence of bone findings (osteopenia, marfanoid habitus, bone deformities)</li> <li>6. History and/or presence of psychiatric symptoms (personality disorder, anxiety, depression, obsessive-compulsive behavior, and psychotic episodes).</li> <li>7. Development             <ol style="list-style-type: none"> <li>a. IQ</li> <li>b. Level of functioning</li> </ol> </li> <li>8. Growth             <ol style="list-style-type: none"> <li>a. Final adult parameters</li> </ol> </li> </ol>
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