

## Arginosuccinic Acid Lyase Deficiency Care Plan (last updated 2/21/09)

| <p><b>Clinical Considerations</b></p> <ul style="list-style-type: none"> <li>• Stabilizing neonate (meds &amp; hemodialysis)</li> <li>• Developmental regression</li> <li>• Progressive liver dysfunction</li> </ul>   | <p><b>Initial labs (diagnostic &amp; baseline)</b></p> <ul style="list-style-type: none"> <li>• Quant plasma amino acids</li> <li>• CMP and ammonia</li> <li>• Consider blood gases, UOA, urine orotic acid, PT/PTT</li> <li>• Consider Argininosuccinate lyase enzyme assay on fibroblasts or sequence analysis</li> </ul>  |           |            |                |               |                |            |                |                |            |  |           |        |
|--|--|-----------|------------|----------------|---------------|----------------|------------|----------------|----------------|------------|--|-----------|--------|
| <p><b>Diet considerations/ treatment</b></p> <ul style="list-style-type: none"> <li>• Low-protein diet</li> <li>• Formula (essential AA)</li> <li>• Medication for hyperammonemia               <ul style="list-style-type: none"> <li>○ Carnitine reported in 1 paper</li> </ul> </li> <li>• Avoidance of fasting</li> <li>• Liver transplant</li> </ul>  | <p><b>Monitoring</b><br/>Quant plasma AA</p> <table border="1" style="margin-left: auto; margin-right: auto; 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;">&gt;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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| 0-6 months   | Every 2 weeks  |           |            |                |               |                |            |                |                |            |  |           |        |
| 6-12 months  | Monthly  |           |            |                |               |                |            |                |                |            |  |           |        |
| 1-6 years  | Every 3 months   |           |            |                |               |                |            |                |                |            |  |           |        |
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| <p><b>Frequency of visits</b></p> <table border="1" style="margin-left: auto; margin-right: auto; 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;">6-24 months</td> <td style="padding: 2px 5px;">Every 3 months</td> </tr> <tr> <td style="padding: 2px 5px;">2-18 years</td> <td style="padding: 2px 5px;">Every 6 months</td> </tr> <tr> <td style="padding: 2px 5px;">&gt;18 years</td> <td style="padding: 2px 5px;">Yearly</td> </tr> </tbody> </table>  | Age  | Frequency | 0-6 months | Every 2 months | 6-24 months   | Every 3 months | 2-18 years | Every 6 months | >18 years      | Yearly     | <p><b>Clinic visit labs</b></p> <ul style="list-style-type: none"> <li>• Comp metabolic panel with LFTs</li> <li>• Consider ammonia, PT, and prealbumin</li> </ul> |           |        |
| Age  | Frequency  |           |            |                |               |                |            |                |                |            |  |           |        |
| 0-6 months   | Every 2 months   |           |            |                |               |                |            |                |                |            |  |           |        |
| 6-24 months  | Every 3 months   |           |            |                |               |                |            |                |                |            |  |           |        |
| 2-18 years   | Every 6 months   |           |            |                |               |                |            |                |                |            |  |           |        |
| >18 years  | Yearly   |           |            |                |               |                |            |                |                |            |  |           |        |
| <p><b>Emergency management</b></p> <ul style="list-style-type: none"> <li>• Immediate 10% (or &gt;) dextrose with salts + intralipids (may need insulin)</li> <li>• If ammonia high, IV meds or dialysis</li> <li>• Track ammonia levels, IV meds (dose), dialysis, central line, intubation, ICU care, days in hospital, coma score</li> <li>• Transfer to peds center if necessary</li> </ul>  | <p><b>Labs to obtain during illness</b></p> <ul style="list-style-type: none"> <li>• Comp metabolic panel</li> <li>• Ammonia</li> <li>• SAA</li> </ul>   |           |            |                |               |                |            |                |                |            |  |           |        |
| <p><b>Other evaluations</b></p> <ul style="list-style-type: none"> <li>• CT /MRI scan at time of Dx if clinically warranted</li> <li>• MRI at 9 y of sooner if clinically warranted</li> <li>• Liver function</li> <li>• Bone health               <ul style="list-style-type: none"> <li>○ DEXA-spine @9y for non-weight bearing &amp; 18y for all others</li> </ul> </li> <li>• Yearly developmental questionnaires (to be completed by parents)</li> <li>• Developmental eval @ 3 &amp; 6y (more frequently if concern of regression)</li> <li>• Neuropsych @ 9y (more frequently if concern of regression)</li> <li>• Psychiatric screening @ 18 y</li> <li>• Referral to Liver and/or Renal as needed</li> <li>• Referral to Neurology if needed</li> <li>• Metabolic dietitian eval (at least yearly)</li> </ul> | <p><b>Yearly labs</b></p> <ul style="list-style-type: none"> <li>• Comp metabolic panel with LFTs</li> <li>• PT/PTT</li> <li>• Consider ammonia</li> <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>  |           |            |                |               |                |            |                |                |            |  |           |        |

# Argininosuccinic Acid Lyase deficiency (ASAL) Care Plan

(last updated 2/21/09)

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| <p><b>Performance Measures</b></p> <ol style="list-style-type: none"> <li>1. Age of diagnosis (both positive NBS and confirmatory SAA)</li> <li>2. Presence of illness at time of diagnosis including lethargy, somnolence, refusal to feed, vomiting, tachypnea, increased neuromuscular tone, spasticity, and ankle clonus.</li> <li>3. Days until ammonia is within treatment range</li> <li>4. Therapy during initial care             <ol style="list-style-type: none"> <li>a. Track IV meds (dose), dialysis, use of central line, intubation, ICU care, days in hospital, and coma score</li> </ol> </li> <li>5. Initial lab studies             <ol style="list-style-type: none"> <li>a. NBS results</li> <li>b. Quant plasma amino acids</li> <li>c. Ammonia</li> <li>d. CMP (w/ LFT's)</li> <li>e. UOA</li> <li>f. Urine orotic acid</li> <li>g. Blood gases</li> <li>h. PT/PTT</li> <li>i. Molecular studies</li> <li>j. Enzymatic studies</li> </ol> </li> <li>6. Monitoring lab studies             <ol style="list-style-type: none"> <li>a. Quant plasma amino acids</li> <li>b. CMP (w/ LFT's)</li> <li>c. Ammonia</li> <li>d. PT/PTT</li> <li>e. Nutrition labs</li> </ol> </li> <li>7. Frequency of clinic contacts and visits (track compliance with visits)</li> <li>8. Total decompensations and hospitalizations.             <ol style="list-style-type: none"> <li>a. Track ICU admissions</li> <li>b. # of days of hospitalization</li> <li>c. # of ER visits</li> <li>d. Track ammonia level, IV meds (dose), dialysis, use of central line, intubation, and coma score</li> </ol> </li> <li>9. Growth parameters (ht, wt, OFC, BMI)</li> <li>10. DEXA results and number of fractures</li> <li>11. Medications (track type and dosage)</li> </ol> | <ol style="list-style-type: none"> <li>12. 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. Medical foods (formula)- (Y/N)</li> <li>e. Modified low-protein products (Y/N)</li> <li>f. Mode (oral, G-tube, bolus/drip, meds only/meds and diet)</li> </ol> </li> <li>13. Transplant (Y/ N)</li> <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. Mortality</li> <li>2. Development             <ol style="list-style-type: none"> <li>a. IQ</li> <li>b. Level of functioning</li> <li>c. Regression</li> </ol> </li> <li>3. History and/or presence of osteopenia</li> <li>4. History and/or presence of seizures</li> <li>5. History and/or presence of neurodegeneration</li> <li>6. History and/or presence of abnormal MRI findings</li> <li>7. History and/or presence of liver dysfunction</li> <li>8. History and/or presence of psychiatric illness</li> <li>9. Outcome of liver transplantation</li> <li>10. Growth             <ol style="list-style-type: none"> <li>a. Final adult parameters</li> </ol> </li> </ol> |
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