

**ACUTE ILLNESS PROTOCOL
UREA CYCLE DISORDERS
THE INFANT/CHILD WITH ARGININOSUCCINATE LYASE DEFICIENCY
(also known as argininosuccinic acidemia)**

PRESENTATION

- Lethargy
- Irritability
- Vomiting
- Ataxia
- Protein avoidance
- Developmental delay
- Failure to thrive
- Hyperammonemia
- Seizures
- Hepatomegaly
- Coma

Apart from arginase deficiency, which usually presents neurologically rather than as a hyperammonemic syndrome, the other urea cycle defects often present in the newborn period with catastrophic hyperammonemia, hepatomegaly, seizures and coma secondary to cerebral edema. Typically OTC and CPS have the most severe presentation but citrullinemia and argininosuccinic acidemia may also present with severe illness. However, all the UCD disorders may present later in life with a severe acute onset or a more chronic course.