

CARE OF PATIENTS WITH METABOLIC DISORDERS IS COMPLEX AND IT IS ESSENTIAL THAT A METABOLIC SPECIALIST BE CONTACTED AS RAPIDLY AS POSSIBLE. THIS PROTOCOL IS ONLY A GUIDELINE AND SHOULD NOT BE USED FOR DEFINITIVE TREATMENT WITHOUT METABOLIC CONSULTATION.

## **ACUTE ILLNESS PROTOCOL**

### **UREA CYCLE DISORDERS**

#### **THE INFANT/CHILD WITH CITRULLINEMIA**

**(also known as argininosuccinate synthetase deficiency)**

#### **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.