

**EMERGENCY PROTOCOL
UREA CYCLE DISORDERS
ARGINASE DEFICIENCY**

DIAGNOSIS

THE DEFINITIVE DIAGNOSIS

If there is uncertainty about the diagnosis, **arginase** can be assayed via red blood cell assay (through the laboratory of Dr. Vivian Shih at the MGH; tel. 617-726 3884/5.)

Differential diagnosis of other metabolic disorders will be assisted by carrying out the **above** tests **PLUS**, assays in the ACUTELY ill child for

- Urinary organic acids/orotic acid/amino acids/acylglycines
- Plasma citrulline/carnitine/acyl carnitines/lactate/pyruvate and where indicated
- CSF Amino acids/lytes/glucose/lactate/pyruvate

The results of these tests will help to direct the differential diagnosis