

## **ACUTE ILLNESS PROTOCOLS**

### **3-HYDROXYMETHYLGLUTARYL-CoA (3-HMG CoA) LYASE DEFICIENCY**

#### ***DIAGNOSIS***

#### **ASSESSMENT**

Assess for dehydration, fever, infection or other stressors that may precipitate an acute metabolic episode. Clinical decompensation can occur rapidly in an infant and may be more gradual in older children.

#### **INVESTIGATIONS**

- Blood glucose (? hypoglycemia)
- pH, blood gases, (? metabolic acidosis)
- electrolytes (? low bicarbonate)
- ammonia (? hyperammonemia)
- urine ketones (? absent or trace)
- urinalysis
- AST, ALT, AP, PT, PTT
- urine organic acids
- culture of blood, throat, urine as indicated

#### **ASSESS BIOCHEMICAL PARAMETERS REGULARLY AND FREQUENTLY WHILE SICK**