

**ACUTE ILLNESS PROTOCOL**  
**FATTY ACID OXIDATION DISORDERS**  
**MEDIUM CHAIN Acyl-CoA DEHYDROGENASE (MCAD) DEFICIENCY**

**CLINICAL PRESENTATION**

- Lethargy
- nausea or vomiting
- hypoglycemia                      with lack or only 'trace' of urinary ketones
- hepatomegaly
- 'Reye' like syndrome
- seizures
- coma
- near/rescued SIDS

Affected infants and children usually present between 3 and 24 months of age particularly when being weaned from nighttime feeds but neonatal cases have been described and adults have become ill after severe exertion (e.g. jogging). The presentation is characterized by **marked lethargy**, often in association with vomiting after a period of fasting. This can progress to hypoglycemic seizures or coma within 1-2 hours of ONSET of symptoms. On occasion seizures or coma may be the presenting sign. Hepatomegaly is usually present. There may, or may not, be a history of a recent viral infection associated with diminished oral intake, or of a similar episode in the past. A history of "recurrent Reye syndrome" should alert you to the possibility of FAODs, as affected children have often been misdiagnosed as having Reye syndrome or 'episodic hypoglycemic coma'; FAODs are responsible for 5-10% of sudden infant death syndrome. Immediate attention and therapy is the key to preventing sudden death.

NOTE that in the acute crises patients can be seriously ill WITHOUT hypoglycemia although typically FAOD crises are associated with hypoglycemia. At these times the urine typically tests 'absent' or 'small' for the presence of ketones. Liver function tests may be mildly elevated; hyperammonemia and hyperuricemia are often present during acute episodes.

**Parents of children with diagnosed metabolic disorders know the early signs of decompensation in THEIR children. Listen to them !!!**