

**ACUTE ILLNESS PROTOCOL
FATTY ACID OXIDATION DISORDERS
LONG CHAIN HYDROXY Acyl-CoA DEHYDROGENASE DEFICIENCY
(LCHADD)**

PRESENTATION

- Hypotonia and weakness
- Lethargy
- Hypoglycemia with absence or 'trace' ketones
- developmental delay
- peripheral neuropathy
- retinitis pigmentosa
- seizures
- hepatomegaly with liver dysfunction (rarely liver failure or cirrhosis)
- coagulopathy
- cardiomyopathy
- 'Reye' like syndrome
- coma
- sudden death

Affected infants and children usually present by 2 years of age. However, neonatal cases do occur. Conversely some patients will not present until adulthood with myoglobinuria and peripheral neuropathy. LCHAD is frequently precipitated by intercurrent illnesses. Children or their sibs affected with fatty acid oxidation disorders have often been misdiagnosed as having Reye syndrome or idiopathic cardiomyopathy; some who have died have also been labeled as SIDS deaths. Such family history should be viewed as suspicious for FAOD.

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.