

ACUTE ILLNESS PROTOCOL ORGANIC ACID DISORDERS

METHYLMALONIC ACIDEMIA

PRESENTATION

- Lethargy
- Vomiting
- Hepatomegaly
- Hypoglycemia
- Hypocalcemia
- Metabolic acidosis
- Hyperammonemia
- Neutropenia

There are two types of presentation, depending on the severity of the metabolic defect. The neonatal form presents within the first days of life with a life-threatening picture of severe lethargy progressing to obtundation. The infantile or late-onset form has a more insidious presentation with failure to thrive, developmental delay, and perhaps other neurologic features such as seizures and spasticity. These children can decompensate acutely during catabolic stress, usually brought on by infection.

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