:: Medium chain fat oxidation disorders

Diseases:

- Medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- 3-hydroxy-3-methylglutaryl-CoA synthase deficiency (HMG-CoA synthase deficiency)
- Carnitine palmitoyl transferase 1A deficiency

The British Inherited Metabolic Disease Group (BIMDG) has published on its website guidelines for the emergency management of patients with inherited metabolic disorders.

Here are the ones for the management of an
Acute decompensation in children and adults with a medium chain fat oxidation disorder.

- See children BIMDG guidelines (Jan 2009)
- See adults BIMDG guidelines (last reviewed in 2012)

Further Information: see the Orphanet Abstracts for MCAD deficiency, HMG-CoA synhtase deficiency and Carnitine palmitoyl transferase 1A deficiency.