:: Defects of ketone metabolism

Diseases:

- Succinyl-CoA acetoacetate transferase deficiency
  - Synonyms: SCOT deficiency ; succinyl CoA oxoacyl CoA transferase deficiency
- Ketoacidosis due to betaketothiolase deficiency
  - Synonyms: Alpha methyl acetoacetyl-CoA thiolase deficiency; MAT deficiency; 3-oxothiolase deficiency; mitochondrial acetoacetyl-CoA thiolase deficiency; T2 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 with a defect of ketone metabolism.

- See children BIMDG guidelines (last reviewed in Feb 2012)

Further Information: see the Orphanet Asbstracts for the SCOT deficiency and the ketoacidosis due to betaketothiolase deficiency.