



British Inherited Metabolic Disease Group

METHYLMALONIC ACIDAEMIA (MMA)  
AND  
PROPIONIC ACIDAEMIA (PA)

COMPLICATIONS OF ACUTE ILLNESS

There are many complications of these disorders and the management can be difficult.

1. **Pancreatitis.** This is probably more common than recognised, partly because it is not easy to diagnose with confidence. It should be suspected if there is abdominal pain, shock out of proportion to other symptoms or hypocalcaemia. Plasma lipase and amylase activity may not be raised, particularly at an early stage.

Abdominal ultrasound may be helpful.

2. **Cardiomyopathy.** This may develop at any time and for reasons not well understood commonly occurs during recovery phase. The symptoms vary from mild (some respiratory distress) to shock and sudden death. Arrhythmias are common and in particular long QTc should be sought (Baumgartner et al 2007). Arrange 2D Echocardiography and electrocardiogram (ECG) if there are any signs of cardio-respiratory problems with regular ECGs when well. All sick patients including those in the recovery phase should be on a cardiac monitor
3. **Stroke-like episodes.** These may occur at any time and frequently of sudden onset. They often involve the basal ganglia and present as a movement disorder. Symptoms may appear at any time, even when the patient is appearing to recover. They may present as hemiplegia or a movement disorder. The aetiology is poorly understood.
4. **Renal disease and failure.** Patients with MMA, but rarely PA, have renal disease. This is initially manifest as a variable renal tubular disorder, often with type IV renal tubular acidosis. Urinary sodium loss may be marked. Renal function almost invariably deteriorates during acute episodes. Fluid and electrolytes need to be managed carefully.

Reference: Baumgartner D, Scholl-Bürgi S, Sass JO, Sperl W et al. Prolonged QTc intervals and decreased left ventricular contractility in patients with propionic acidemia. *J Pediatr.* 2007 ;**150**(2):192-7,