Methylmalonyl-CoA Epimerase Deficiency Case: Mimicking Ketolysis Defect

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Background
Methylmalonyl CoA epimerase (MCE, another name with methylmalonyl-CoA racemase) catalyzes the transformation of (S)-methylmalonyl-CoA to (R)-methylmalonyl-CoA in the propionyl-CoA to succinyl-CoA pathway. Therefore, MCE deficiency (OMIM 251120) has been reported in very few cases.

Case Report
• The male patient was born from consanguineous parents following an uneventful pregnancy and delivery.
• He had typical neuromotor milestones.
• However, when he was three years old, he had severe metabolic ketoacidosis at his first metabolic decompensation.
• His blood β-ketone, in fresh capillary whole blood from the fingertip, 5.2 mmol/l (N<0.6) was elevated.
• Organic acid profiles were dominated by increased 3 hydroxybutyrate, and acylcarnitine profiles showed marked C3 (propionyl carnitine) elevation.
• ACAT1 deficiency in ketolysis defects was initially suspected, but it was subsequently noted that methylmalonic acid was mildly but persistently elevated in urine.
• MCEE gene analysis was homozygous for p.R47X (c.139HC>T) nonsense.

Discussion
• The clinical spectrum of MCE deficiency ranges from asymptomatic to life-threatening metabolic decompensation attacks.
• This report of a patient diagnosed with MCE deficiency with acute metabolic ketoacidosis attacks and moderate MMA-uria persists in periods without decompensation.
• In addition, this report provides a new phenotype of the clinical and biochemical characterization of MCE deficiency.
References


