**Abstract:**
We describe a 4-day old neonate, seventeenth reported patient with carbonic anhydrase VA deficiency, who presented with hyperammonaemia, lactic acidosis and ketoacidosis but without hypoglycaemia and without elevation of the organic acid metabolites expected from dysfunction of bicarbonate-dependent mitochondrial carboxylase enzymes. The absence of elevated propionyl-CoA and 3-methylcrotonyl-CoA metabolites is in contrast to some earlier described patients and highlights the importance of considering CAVA deficiency in neonates presenting with isolated hyperammonaemia, lactic acidosis and ketoacidosis.

**Introduction:**
Carbonic Anhydrases are a family of zinc metalloenzymes that catalyse the reversible hydration of CO₂ to bicarbonate.

As bicarbonate cannot cross the mitochondrial membrane, carbonic anhydrase VA (CAVA) isoenzyme is essential in accelerating the conversion of CO₂ to bicarbonate within the mitochondria.

Bicarbonate and ammonia are substrates for carbamoyl phosphate synthetase 1 in the urea cycle (Figure 1). Bicarbonate is also an important substrate for three biotin-dependent intramitochondrial carboxylase enzymes: Pyruvate carboxylase converts pyruvate to oxaloacetate, which promotes gluconeogenesis, tricarboxylic acid cycle anaplerosis and formation of aspartate (Figure 2).

Propionyl-CoA carboxylase and 3-methylcrotonyl-CoA carboxylase are involved in the metabolism of branched chain amino acids valine, isoleucine, and leucine (Figure 3, 4).

**References:**