# A Rare Case of β-Ketothiolase Deficiency

B. Modh[1], G. Pathak[2], A. Chauhan[3]

**ABSTRACT**

We are reporting a case of β-ketothiolase deficiency, a rare disorder of amino acid metabolism. A 10 month old child presented with complaints of vomiting, convulsions, fever and altered sensorium that on investigations showed metabolic acidosis, hyperammonemia and ketosis. Gas chromatography/ mass spectroscopic examination was suggestive of β-ketothiolase deficiency.

**Key words:** β-ketothiolase deficiency, metabolic acidosis, ketosis

**Authors:**

[1]Resident, [2]Professor and Head of Unit, [3]Assistant professor, Dept. of Pediatrics, B.J.Medical College, Ahmedabad.