

Muscle b-enolase deficiency presenting with different kinetic profiles in muscle tissue.

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Background : Muscle b-enolase deficiency is a very rare inherited metabolic myopathy caused by an enzymatic defect of distal glycolysis. We aim to analyse the kinetic profile of total enolase activity in muscle tissue of patients with this condition.

Methods or Patients or Materials: Case series.

Results: A 16 years old male Asiatic teenager presented with recurrent rhabdomyolysis (highest CK: 193,000). The first episode happened at the age of 14 years. His baseline CK was normal. Muscle biopsy showed minimal changes. A forearm test revealed low lactate and ammonia production. The second patient is a 28 years old Turkish patient who presented with exercise related muscle pain and rhabdomyolysis (highest CK 75,000 IU/L) since childhood. His baseline CK was also normal. A forearm test revealed low lactate production. Biochemical studies on muscle tissue showed diminished total enolase activities but differing kinetic profiles. Genetic test confirmed homozygous *ENO3* mutations in both patients.

Conclusion: Muscle b-enolase deficiency is a rare cause of rhabdomyolysis. Here we report two cases with different kinetic profiles of total enolase activity in muscle tissue.