Clinical Genetics

AB055. *HADHB* mutations in a child with suspected metabolic myopathy

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Background: Metabolic myopathies should be considered in patients with recurrent rhabdomyolysis, one episode triggered by prolonged fasting, fever or infection, symptoms such as myalgia or exercise intolerance, and otherwise unexplained serum CK elevation. The defects of long-chain fatty acid beta-oxidation are one of common causes of metabolic myopathy in childhood. With the exception of carnitine palmitoyltransferase II (CPT II) deficiency, the most common cause, there is a rather rare mitochondrial trifunctional protein (MTP) deficiency.

Methods: We studied total 11 patients with suspected metabolic myopathy in Seoul National University Children's Hospital between April 2003 and December 2016. The causes were not revealed by metabolic work-up including acylcarnitine profile. Sanger sequencing of *CPT2* and *HADHB* genes were performed.

Results: One patient was diagnosed with MTP deficiency. Mutation analysis of *HADHB* revealed compound heterozygosity for c.340A>G (p.N114D) and c.1364T>G (p.V455G) in one patient. Sequencing of the *CPT2* identified no causative variants. The proband, a 3-year-old male, presented with recurrent rhabdomyolysis and exercise intolerance. The result of tandem mass spectrometry revealed normal finding even during the attacks.

Conclusions: The defects of lipid metabolism such as MTP deficiency should be considered in patients with suspected metabolic myopathy. This report shows that acylcarnitine profile can be normal even during the attacks, although biochemical analysis such as plasma carnitine and acylcarnitine profile is usually valuable as screening method. Therefore genetic analysis searching for mutations in *CPT2* and *HADHB* genes should be performed in all the patients with otherwise unexplained metabolic myopathy.

Keywords: Metabolic myopathy; *HADHB* gene; mitochondrial trifunctional protein (MTP) deficiency

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