Coexistence of Megaconial Congenital Muscular Dystrophy and Cystinuria: Mimicking Hypotonia–Cystinuria Syndrome

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Objectives:

Hypotonia-cystinuria syndrome (HCS) is characterized by cystinuria type A, developmental delay, growth hormone deficiency, and hypotonia findings. Here in, we present a sixteen-month-old male patient admitted with the clinical findings of hypotonia-cystinuria syndrome and diagnosed with megaconial congenital muscular dystrophy and cystinuria.

Case presentation:

A 16-month-old male presented with hypotonia and growth retardation since the age of 2 months. On physical examination, developmental delay, mild hypotonia, and growth retardation were present. In laboratory studies, mild elevated serum creatinine kinase and high dibasic amino acid in quantitative urine amino acid analysis were found. Therefore, genetic analysis was performed for the differential diagnosis of HSC. No deletion was detected at 2p21. Whole exome sequencing analysis was performed to explain clinical and laboratory findings. As a result, heterozygous mutation of c.1266_1267delGT in SLC7A9 gene and a novel homozygous c.225-2A>T pathogenic variant in CHKB gene were found. Co-occurrence of megaconial congenital muscular dystrophy and cystinuria which mimicked hypotonia-cystinuria syndrome was detected.

Conclusion:

Different rare diseases, which are more common in countries with a high incidence of consanguineous marriage, can sometimes coexist in the same person. Whole exome sequencing analysis is a reliable diagnostic method to reveal coexistence of rare disorders.