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Compound heterozygous variants in GOSR2 associated with congenital muscular dystrophy: A case report

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Abstract

The homozygous missense variant in the GOSR2 gene (c.430G>T) is known to be associated with progressive myoclonic epilepsy (PME). The clinical presentation of GOSR2-related PME involves the development of ataxia, seizures, scoliosis, areflexia, and mildly elevated creatine kinase. Recently, it has been suggested that some compound heterozygous variants in GOSR2 are associated with a predominant muscular dystrophy phenotype. Here we report a case of a now 22 month old female who presented with congenital hypotonia and persistently elevated creatine kinase levels. Whole exome sequencing showed pathogenic compound heterozygous variants in GOSR2 (c.430G>T and c.82C>T). This case contributes to the expanding clinical spectrum of GOSR2 variants with PME representing the milder end and congenital muscular dystrophy representing the more severe end of the spectrum.

Keywords: GOSR2; PME; dystroglycanopathy; muscular dystrophy; progressive myoclonic epilepsies.

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