

Reporting One Very Rare Pathogenic Variation c.1106G>A in POMT2 Gene

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PMID: 32494558  PMCID: PMC7263986  DOI: 10.5582/irdr.2020.03013

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Abstract

Dystroglycan (DG) is a major cell membrane glycoprotein, which is encoded by the DAG1 gene. α-DG is one of DG subunits, belongs to O-mannosylated protein of mammals and was identified in brain, peripheral nerves and muscle. Dystroglycanopathies are a group of heterogeneous congenital muscular dystrophies, which can result from defective α-DG mannosylation. First line of α-DG glycosylation is catalyzed by protein O-mannosyltransferase family (PMT). In this study, the mutation was identified in the POMT2 gene, which encodes O-mannosyltransferase 2 protein and its mutations can be contributed to dystroglycanopathies. A very rare missense mutation in the POMT2 gene (NM_013382: exon9: c. 1106G>A) was identified by next generation sequencing (NGS) and was subsequently confirmed using Sanger sequencing in both affected siblings. There was no report of this mutation in the literature, therefore, the significance was uncertain. Our findings confirmed the pathogenicity of mutation and expanded the mutation spectrum of POMT2, which will be helpful in further molecular evaluations of muscular diseases.

Keywords: POMT2; alpha-dystroglycan; dystroglycanopathy; rare mutation.

2020, International Research and Cooperation Association for Bio & Socio - Sciences Advancement.

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