ScienceDirect

Neuromuscular Disorders
Volume 33, Supplement 1, October 2023, Pages S116-S117

# P293 Clinical and genetic spectrum of sarcoglycanopathies in a cohort of Turkish patients with a possible founder variation

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https://doi.org/10.1016/j.nmd.2023.07.203
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Sarcoglycanopathies consist of four subtypes of autosomal-recessive limb-girdle-muscular-dystrophies(LGMD2C/R5, LGMD2D/R3, LGMD2E/R4, LGMD2F/R6) caused by mutations in the SGCG, SGCA, SGCB, and SGCD genes, respectively. Very little is known about the clinical and genetic characteristics of sarcoglycanopathies in Turkish patients. Twenty patients with molecularly confirmed sarcoglycanopathies, including 11 with LGMD2D/R3, 3 with LGMD2E/R4, 1 with LGMD2F/R6 and 7 with LGMD2C/R5 are included. As reported in many countries, LGMD2D/R3 is the most prevalent form in our country, and followed by LGMD2C/R5, LGMD2E/R4, and LGMD2F/R6. The mean age of molecular genetic diagnosis is 17,4 and the mean age at onset of symptoms were 6,9. Severe childhood-onset LGMD patients have a higher probability of obtaining a molecular diagnosis than adult-onset LGMD patients. The symptoms at the time of onset were varied with different degrees of proximal lower limb weakness, early fatigue, frequent falls, gait abnormalities, delayed motor milestones, exercise intolerance. All of the variants were homozygous and the most common pathogenic variant was SGCA:c.850C>T(4 patients). A probable founder SGCG gene-variant(c.392A>G) that had been classified as variant-of-uncertain-significance before, was detected in 4 patients whose diagnosis was confirmed with muscle biopsy. With the rapid developments of gene therapies in sarcoglicanopathies, the molecular diagnosis of the patients is getting essential.

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