CO158

Loss of Ambulation in Patients with Limb-Girdle Muscular Dystrophy Sarcoglycanopathy Subtypes: A Systematic Review

Supplementary Appendix

Antoinette Cheung, MPH,[1] Sharanya Murty, PhD,[2] André Müller-York, MD,[2] Lavanya Sudharshan, MS,[2] Maria Tinajero, MSc,[1] Shelagh M. Szabo, MSc[1]

[1] Broadstreet HEOR, Vancouver BC Canada, [2] Sarepta Therapeutics, Inc., Cambridge, MA USA

References

1. Limb-Girdle Muscular Dystrophies. 2019; https://rarediseases.org/.
2. Sandonà D, Betto R. Sarcoglycanopathies: molecular pathogenesis and therapeutic prospects. *Expert Rev Mol Med*. 2009;11:e28. doi:10.1017/S1462399409001203
3. Audhya IF, Cheung A, Szabo SM, Flint E, Weihl CC, Gooch KL. Progression to Loss of Ambulation Among Patients with Autosomal Recessive Limb-girdle Muscular Dystrophy: A Systematic Review. *J Neuromuscul Dis.* 2022;9(4):477-492. doi:10.3233/JND-210771
4. Cheung A, Audhya IF, Szabo SM, Friesen M, Weihl CC, Gooch KL. Patterns of Clinical Progression Among Patients With Autosomal Recessive Limb-Girdle Muscular Dystrophy: A Systematic Review. *J Clin Neuromuscul Dis.* 2023;25(2):65-80. doi:10.1097/CND.0000000000000461
5. Alavi A, Esmaeili S, Nilipour Y, et al. LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. *J Neurogenet.* 2017;31(3):161-169. doi:10.1080/01677063.2017.1346093*
6. Ginjaar HB, van der Kooi AJ, Ceelie H, et al. Sarcoglycanopathies in Dutch patients with autosomal recessive limb girdle muscular dystrophy. *J Neurol.* 2000;247(7):524-529. doi:10.1007/s004150070151*
7. Lodi R, Muntoni F, Taylor J, et al. Correlative MR imaging and 31P-MR spectroscopy study in sarcoglycan deficient limb girdle muscular dystrophy. *Neuromuscul Disord.* 1997;7(8):505-511. doi:10.1016/s0960-8966(97)00108-9*
8. Pegoraro V, Angelini C. Circulating miR-206 as a Biomarker for Patients Affected by Severe Limb Girdle Muscle Dystrophies. *Genes (Basel).* 2021;12(1):85. doi:10.3390/genes12010085*
9. Rosenbloom E, Anziska Y, Juan CS, Stickevers S. Three Siblings with Varying Phenotypic Expression of Limb Girdle Muscular Dystrophy 2C: A Case Series [abstract]. AAPM&R Meeting Abstracts. 2021; 13(1)*
10. Ten Dam L, de Visser M, Ginjaar IB, van Duyvenvoorde HA, van Koningsbruggen S, van der Kooi AJ. Elucidation of the Genetic Cause in Dutch Limb Girdle Muscular Dystrophy Families: A 27-Year's Journey. *J Neuromuscul Dis*. 2021;8(2):261-272. doi:10.3233/JND-200585*
11. Xie Z, Hou Y, Yu M, et al. Clinical and genetic spectrum of sarcoglycanopathies in a large cohort of Chinese patients. *Orphanet J Rare Dis.* 2019;14(1):43. Published 2019 Feb 14. doi:10.1186/s13023-019-1021-9*
12. Liang WC, Jong YJ, Wang CH, et al. Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with limb-girdle muscular dystrophy. *Orphanet J Rare Dis.* 2020;15(1):160. doi:10.1186/s13023-020-01445-1*
13. Tétreault M, Srour M, Allyson J, et al. Founder mutation for α-sarcoglycan-LGMD2D in a Magdalen Islands Acadian cluster. *Can J Neurol Sci.* 2011;38(5):747-752. doi:10.1017/s0317167100054135*
14. Bönnemann CG, Passos-Bueno MR, McNally EM, et al. Genomic screening for beta-sarcoglycan gene mutations: missense mutations may cause severe limb-girdle muscular dystrophy type 2E (LGMD 2E). *Hum Mol Genet.* 1996;5(12):1953-1961. doi:10.1093/hmg/5.12.1953*
15. Fanin M, Melacini P, Boito C, Pegoraro E, Angelini C. LGMD2E patients risk developing dilated cardiomyopathy. *Neuromuscul Disord.* 2003;13(4):303-309. doi:10.1016/s0960-8966(02)00280-8*
16. Marchetti GB, Valenti L, Torrente Y. Clinical Determinants of Disease Progression in Patients With Beta-Sarcoglycan Gene Mutations. *Front Neurol.* 2021;12:657949. doi:10.3389/fneur.2021.657949*
17. Pashun RA, Azari BM, Achar A, et al. Intramyocardial Fat in Family With Limb-Girdle Muscular Dystrophy Type 2E Cardiomyopathy and Sudden Cardiac Death. *Circ Cardiovasc Imaging.* 2020;13(7):e010104. doi:10.1161/CIRCIMAGING.119.010104*
18. Semplicini C, Vissing J, Dahlqvist JR, et al. Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*. 2015;84(17):1772-1781. doi:10.1212/WNL.0000000000001519*
19. Tariq M, Latif M, Inam M, et al. Whole exome sequencing reveals a homozygous SGCB variant in a Pakhtun family with limb girdle muscular dystrophy (LGMDR4) phenotype. *Gene Rep*. 2021;22 (101014)*
20. Alonso-Pérez J, González-Quereda L, Bruno C, et al. Clinical and genetic spectrum of a large cohort of patients with δ-sarcoglycan muscular dystrophy. *Brain.* 2022;145(2):596-606. doi:10.1093/brain/awab301*
21. Guimarães-Costa R, Fernández-Eulate G, Wahbi K, et al. Clinical correlations and long-term follow-up in 100 patients with sarcoglycanopathies. *Eur J Neurol.* 2021;28(2):660-669. doi:10.1111/ene.14592
22. Alonso-Pérez J, González-Quereda L, Bello L, et al. New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. *Brain*. 2020;143(9):2696-2708. doi:10.1093/brain/awaa228

***Reported patient-level data on ambulatory status among patients with sarcoglycanopathies**