

Phenotype Presentation and Molecular Diagnostic Yield in Non-5q Spinal Muscular Atrophy

Neurol Genet 2024;10:e200123. doi:10.1212/NXG.0000000000200123

In the Research Article “Phenotype Presentation and Molecular Diagnostic Yield in Non-5q Spinal Muscular Atrophy” by Fernández-Eulate et al.,¹ the last 3 sentences of the Results section’s final paragraph should read as follows:

“Direct sequencing of the *COL6A3* gene found a previously reported homozygous c.7447A>G variant²² as well as a previously undescribed heterozygous missense variant (c.5867A>G), which could possibly acts as a modulator of the disease. A sibling, who presents a similar clinical phenotype and a predominant neurogenic ENMG and muscle biopsy carried the same variants. Their mother was heterozygous for the c.7447A>G variant and the father was deceased.”

The authors regret the errors.

Reference

1. Fernández-Eulate G, Theuriet M, Record CJ, et al. Phenotype presentation and molecular diagnostic yield in non-5q spinal muscular atrophy. *Neurol Genet.* 2023;9(4):e200087. doi:10.1212/NXG.0000000000200087