

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

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In the Research Article “Phenotype Presentation and Molecular Diagnostic Yield in Non-5q Spinal Muscular Atrophy” by Fernández-Eulate et al.,¹ the first paragraph under the Other Non-5q SMA–Associated Genes section should read as follows to avoid any misunderstanding:

Other variants in genes implicated in HMNs were found. In 2 SP-SMA patients with lower-limb distal involvement, scapula alata, and a disease onset in adult life, heterozygous HSPB1 variants were found. Two patients (SP-SMA and P-SMA) carried heterozygous variants in the VCP and one (SP-SMA) a homozygous variant in the VRK1 gene, all 3 with an adult disease onset. A previously reported patient with an adult-onset PD-SMA carried a heterozygous variant in the MORC2 gene.¹⁸ One patient with an infantile-onset PD-SMA carried a homozygous pathogenic variant in the DNAJB2 gene. The specific genetic variants can be seen in eTable 1 (links.lww.com/NXG/A614).

Additionally, in eTable 1, the first variant of patient number 11 has been corrected to “(p.Arg598Cys)”, and the first variant of patient number 56 has been corrected to “Homozygous c.961C>T (p.Arg321Cys).” The updated eTable 1 can be accessed at links.lww.com/NXG/A614.

The authors regret the errors.

Reference

1. Fernández-Eulate G, Theuriet J, 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.000000000200087