Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3

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Patients 3-1 and 3-2 (siblings) were tested at GeneDx. They were compound heterozygous for p.Arg1472Glu and c.3434del p.Lys1145fs*7. However c.3434del p.Lys1145fs*7 was noted as p.Leu1145fs*7 in Table S1 and the supplemental text. This has now been corrected, and the authors regret the error.

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