

Whole exome sequencing of a patient with suspected mitochondrial myopathy reveals novel compound heterozygous variants in *RYR1*

In Blackburn et al. (2017), the novel frameshift deletion variant that appeared throughout the article was published as:

“c.4485_4500del, p.Tyr1495X”

It should have read as:

“c.4485_4500del, p.Trp1495X”

We also referred to a nonsense variant reported in Klein et al. (2012) (<https://doi.org/10.1002/humu.22136>) in their supplemental Table S1b as ‘c.4485G>A; p.Tyr1495X,’ that results in an equivalent protein truncation. This variant should also be reported as c.4485G>A, p.Trp1495X.

We apologize for this error.

REFERENCE

Blackburn, P. R., Selcen, D., Gass, J. M., Jackson, J. L., Macklin, S., Cousin, M. A., ... Atwal, P. S. (2017). Whole exome sequencing of a patient with suspected mitochondrial myopathy reveals novel compound heterozygous variants in *RYR1*. *Molecular Genetics & Genomic Medicine*, 5(3), 295–302.