



Correction: Interpretation of mitochondrial tRNA variants

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The original version of this Article contained an error in the spelling of "Criteria for mt-tRNA variants" in the top right column header of Table 2, which was incorrectly given as "mt-mRNA." This has now been corrected in both the PDF and HTML versions of the Article.

Table 2 Criteria for the classification of benign variants.

Evidence	ACMG criteria	Criteria for mt-tRNA variants
Stand- alone	BA1 Allele frequency is above 5% in Exome Sequencing Project, 1000 Genomes, or ExAC.	BA1 Top-level haplogroup defining variants.
Strong	BS1 Allele frequency is greater than expected for disorder.	BS1 Reported in public databases (e.g., MitoMap or mtDB) or literatures as polymorphism.
	BS2 Observed in healthy adult individual for a recessive (homozygous), dominant (heterozygous), or X-linked (hemizygous) disorder with full penetrance expected at an early age.	BS2 Found homoplasmic in more than three unrelated healthy adults.
	BS3 Well-established in vitro or in vivo functional studies shows no damaging effect on protein function or splicing.	BS3 Not applicable.
	BS4 Lack of segregation in affected members of a family.	BS4 Homoplasmy in both probands and at least 2 asymptomatic matrilineal family members.
Supporting	BP1 Missense variant in a gene for which primarily truncating variants are known to cause disease.	BP1 Not applicable.
	BP2 Observed in <i>trans</i> with a pathogenic variant for a fully penetrant dominant gene/disorder; or observed in <i>cis</i> with a pathogenic variant in any inheritance pattern.	BP2 Not applicable.
	BP3 In-frame deletions/insertions in a repetitive region without a known function.	BP3 Not applicable.
	BP4 Multiple lines of computational evidence suggest no impact on gene or gene product.	BP4 The MitoTIP prediction score is <10.
	BP5 Variant found in a case with an alternate molecular basis for disease.	BP5 In the presence of a known pathogenic genetic cause unless there is evidence of more than one disease and clinically explained.
	BP6 Reputable source recently reports variant as benign but the evidence is not available to the laboratory to perform an independent evaluation.	BP6 Found to be homoplasmic more than once in asymptomatic adults in private reputable laboratory databases.
	BP7 A synonymous (silent) variant for which splicing prediction algorithms predict no impact to the splice consensus sequence nor the creation of a new splice site AND the nucleotide is not highly conserved.	BP7 Not applicable.

The ACMG criteria are listed on the left column and the new criteria for mt-tRNA variants are on the right.

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