

Variant: *NM_000277.2(PAH):c.500A>G (p.Asn167Ser)*

Version: 1.0

CA229585 [↗](#)

102703 (ClinVar) [↗](#)

Gene: PAH ([HGNC:5053](#))

Condition: phenylketonuria ([MONDO:0009861](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 33e1f5d5-20ab-45d2-8dcd-4eba431a8642

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HGVS expressions

NM_000277.2:c.500A>G

NM_000277.2(PAH):c.500A>G (p.Asn167Ser)

NC_000012.12:g.102866605T>C

CM000674.2:g.102866605T>C

NC_000012.11:g.103260383T>C

CM000674.1:g.103260383T>C

NC_000012.10:g.101784513T>C

NG_008690.1:g.55998A>G

NG_008690.2:g.96806A>G

NM_000277.1:c.500A>G

NM_001354304.1:c.500A>G

NM_000277.3:c.500A>G

ENST00000307000.7:c.485A>G

ENST00000549111.5:n.596A>G

ENST00000551988.5:n.530+10857A>G

ENST00000553106.5:c.500A>G

Uncertain Significance

Met criteria codes **2**

PP4 BS1

Not Met criteria codes **1**

PP3

Evidence Links **1**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

PAH-specific ACMG/AMP criteria applied: PP4: N167S was observed in 1 case (genotype N167S/-) with benign persistent hyperphenylalaninemia (200-600 uM). (PMID:11385716); BS1: gnomAD MAF: 0.01461. In summary this variant meets criteria to be classified as uncertain significance for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PP4, BS1).

Met criteria codes

PP4	✓	N167S was observed in 1 case (genotype N167S/-) with benign persistent hyperphenylalaninemia (200-600 uM).
		N167S was observed in 1 case (genotype N167S/-) with benign persistent hyperphenylalaninemia. Benign persistent hyperphenylalaninemia was diagnosed in cases with the untreated PHE level of 4-9.9 mg/dl (200-600 μM). PubMed:11385716
BS1	✓	gnomAD MAF: 0.01461

Not Met criteria codes

PP3	✗	Conflicting interpretations of pathogenicity: benign and tolerated in SIFT, PP2. Deleterious in MutationTaster
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Curation History [↗](#)

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