

*Variant: NM\_000277.2(PAH):c.500A>G (p.Asn167Ser)*

CA229585 [↗](#)

102703 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal recessive inheritance

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

**Approved on:** 2018-08-13

**Published on:** 2018-12-10

### *HGVS expressions*

**NM\_000277.2(PAH):c.500A>G (p.Asn167Ser)**

**NM\_000277.2:c.500A>G**

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

Curation History

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