

## 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

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

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

**Published on:** 2019-04-06

### *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

<b>PP4</b>	✓	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). <a href="#">PubMed:11385716</a>
<b>BS1</b>	✓	gnomAD MAF: 0.01461

#### Not Met criteria codes

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



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See Report	Preferred Variant Title	Classification ⓘ	Condition	Published Date	Version ⓘ	Criteria Specification	Gene
<a href="#">View</a>	NM_000277.2(PAH):c.500A>G (p.Asn16...	<b>Uncertain Significance</b>	<a href="#">Phenylketonuria</a>	2019-04-06	1.0	-	<a href="#">PAH</a>

Showing 1 to 1 of 1 rows

