The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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
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

BS1 PP4

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

BS1	✔	gnomAD MAF: 0.01461
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

Not Met criteria codes

PP3	✖	Conflicting interpretations of pathogenicity: benign and tolerated in SIFT, PP2. Deleterious in MutationTaster

Curation History

Showing 1 to 1 of 1 rows