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

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