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Variant: NM_000277.3(PAH):c.1048T>A (p.Ser350Thr)

Version: 1.0

CA229304

102494 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: b924ccb3-a7dd-467c-a8d4-f7f78a645076
Approved on: 2021/2/16
Published on: 2021/6/9

HGVS expressions

NM_000277.3:c.1048T>A
NM_000277.3(PAH):c.1048T>A (p.Ser350Thr)
NC_000012.12:g.102844353A>T
CM000674.2:g.102844353A>T
NC_000012.11:g.103238131A>T
CM000674.1:g.103238131A>T
NC_000012.10:g.101762261A>T
NG_008690.1:g.78250T>A
NG_008690.2:g.119058T>A
ENST00000553106.6:c.1048T>A
ENST00000307000.7:c.1033T>A
ENST00000549247.6:n.807T>A
ENST00000551114.2:n.710T>A
ENST00000553106.5:c.1048T>A
ENST00000635477.1:c.152T>A
ENST00000635528.1:n.563T>A
NM_000277.1:c.1048T>A
NM_000277.2:c.1048T>A
NM_001354304.1:c.1048T>A
NM_001354304.2:c.1048T>A

Likely Pathogenic

Met criteria codes 4

PM5 PM2 PP3 PP4_Moderate

Evidence Links 1

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.1048T>A (p.Ser350Thr) variant in PAH has been reported in 1 individual with classic PKU and BH4 deficiency excluded (PMID: 9634518). This variant is absent in population databases. Computational evidence supports a deleterious effect. A different pathogenic

missense change (p.Ser350Tyr) has been seen at the same amino acid. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM5, PP3.

Met criteria codes

PM5	✔	P350Y classified as pathogenic on ClinVar (Var ID: 102495)
PM2	✔	Absent in controls in ExAC,1000 Genomes or ESP.
PP3	✔	Pedicted deleterious by SIFT, Polyphen, MutationTaster
PP4_Moderate	✔	Identified in one patient with Classic PKU, BH4 deficiency excluded. PMID: 9634518
		Identified in one patient with Classic PKU, but listed as unclassified. **PubMed:9634518**

Curation History

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