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Variant: NM_000277.2(PAH):c.164T>C (p.Phe55Ser)

CA267639

120266 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: cd7b08c0-d1ff-4dc6-95f5-142a0fb35f8b
Approved on: 2018-12-10
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HGVS expressions

NM_000277.2:c.164T>C
NM_000277.2(PAH):c.164T>C (p.Phe55Ser)
NC_000012.12:g.102912795A>G
CM000674.2:g.102912795A>G
NC_000012.11:g.103306573A>G
CM000674.1:g.103306573A>G
NC_000012.10:g.101830703A>G
NG_008690.1:g.9808T>C
NG_008690.2:g.50616T>C
NM_000277.1:c.164T>C
NM_001354304.1:c.164T>C
NM_000277.3:c.164T>C
ENST00000307000.7:c.149T>C
ENST00000546844.1:c.164T>C
ENST00000548677.2:n.251T>C
ENST00000548928.1:n.86T>C
ENST00000549111.5:n.260T>C
ENST00000550978.6:n.148T>C
ENST00000551337.5:c.164T>C
ENST00000551988.5:n.253T>C
ENST00000553106.5:c.164T>C
ENST00000635500.1:n.132T>C

Likely Pathogenic

Met criteria codes 4
PM3 PM2 PP3 PP4

Not Met criteria codes 1
PM5

Evidence Links 1

Expert Panel
Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.164T>C (p.Phe55Ser) variant in PAH is reported in 1 French patient with classic PKU. BH4 deficiency was not assessed. It was detected with a known pathogenic variant c.1066-11G>A (PMID: 26666653). This variant is absent from ExAC, gnomAD, 1000G, and ESP. A deleterious effect is predicted in SIFT, Polyphen-2, MutationTaster, and REVEL=0.96. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP4, PP3.

Met criteria codes

PM3	✔	Detected with c.1066-11G>A, PMID: 26666653 Patient genotype: c. [164T>C]; [1066-11G>A]. PubMed:26666653
PM2	✔	Absent from ExAC, gnomAD, 1000G, ESP
PP3	✔	Deleterious effect predicted in SIFT, Polyphen2, MutationTaster. REVEL=0.96
PP4	✔	Identified in 1 French patient with classic PKU. BH4 deficiency not assessed. PMID: 26666653 Phe55Ser identified in 1 patient with classic PKU. A total of 364 hyperphenylalaninemic patients, including 42 siblings, were investigated. They represented mixed origin people from 20 French Centers for Inborn Errors of Metabolism. According to their blood Phenylalanine concentration at time of diagnosis, these patients were assigned to one of the three phenotype categories. classical phenylketonuria (cPKU, Phe > 1200 µmol/L). PubMed:26666653

Not Met criteria codes

PM5	✖	F55L P/LP

Curation History

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