

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

Published on: 2018-12-10

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**

PP3

PP4

PM2

PM3

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

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 			
PM2			Absent from ExAC, gnomAD, 1000G, ESP
PM3			Detected with c.1066-11G>A, PMID: 26666653
Patient genotype: c. [164T>C]; [1066-11G>A]. PubMed:26666653 			

Not Met criteria codes

PM5			F55L P/LP
-----	---	---	-----------

Curation History



 

 

Showing 1 to 1 of 1 rows

--

