

Variant: *NM_000277.2(PAH):c.884C>G (p.Ser295Ter)*

Version: 1.0

CA229834 [↗](#)

102884 (ClinVar) [↗](#)

Gene: PAH ([HGNC:5053](#))

Condition: phenylketonuria ([MONDO:0009861](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 6d125790-217d-4bb8-aeab-b6ee4f35be49

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HGVS expressions

NM_000277.2(PAH):c.884C>G (p.Ser295Ter)

NM_000277.2:c.884C>G

NC_000012.12:g.102851715G>C

CM000674.2:g.102851715G>C

NC_000012.11:g.103245493G>C

CM000674.1:g.103245493G>C

NC_000012.10:g.101769623G>C

NG_008690.1:g.70888C>G

NG_008690.2:g.111696C>G

ENST00000553106.6:c.884C>G

ENST00000307000.7:c.869C>G

ENST00000549247.6:n.643C>G

ENST00000551114.2:n.546C>G

ENST00000553106.5:c.884C>G

ENST00000635477.1:c.45C>G

NM_000277.1:c.884C>G

NM_001354304.1:c.884C>G

NM_000277.3:c.884C>G

NM_001354304.2:c.884C>G

Pathogenic

Met criteria codes **3**

PVS1 **PP4** **PM2**

Not Met criteria codes **1**

PM3

Evidence Links **1**

Expert Panel

[Phenylketonuria VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.884C>G (p.S295*) variant in PAH has been reported in 1 individual with PKU, however without indication of complete genotype (PMID:10394930). This variant is absent in population databases, including 1000 Genomes, Exome Sequencing Project (ESP) and the

Genome Aggregation database (gnomAD). This is a nonsense variant in exon 8 of 13 in PAH, predicted to undergo nonsense mediated decay with the truncated region critical to protein function. Overall this variant meets criteria to be classified as pathogenic for PAH. ACMG/AMP criteria applied: PM2, PVS1, PP4.

Met criteria codes

PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			Reported in a phenyketonuria cohort in an allele of Arabian origin. Reported in a phenyketonuria cohort in an allele of Arabian origin. PubMed:10394930 
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

PM3			Reported in a phenyketonuria cohort in an allele of Arabian origin. The genotype of the affected individual was not reported. PubMed:10394930 
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Curation History

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