

Variant: *NM_001354304.2:c.209C>T*

Version: 1.0

CA16020747 [↗](#)

987767 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 2f26a145-c76e-4696-9509-47d4ce44f81a

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HGVS expressions

NM_001354304.2:c.209C>T

NC_000012.12:g.102894878G>A

CM000674.2:g.102894878G>A

NC_000012.11:g.103288656G>A

CM000674.1:g.103288656G>A

NC_000012.10:g.101812786G>A

NG_008690.1:g.27725C>T

NG_008690.2:g.68533C>T

ENST00000553106.6:c.209C>T

ENST00000307000.7:c.194C>T

ENST00000546844.1:c.209C>T

ENST00000548677.2:n.296C>T

ENST00000548928.1:n.131C>T

ENST00000549111.5:n.305C>T

ENST00000550978.6:c.193C>T

ENST00000551337.5:c.209C>T

ENST00000551988.5:n.298C>T

ENST00000553106.5:c.209C>T

ENST00000635500.1:n.177C>T

NM_000277.1:c.209C>T

NM_000277.2:c.209C>T

NM_001354304.1:c.209C>T

NM_000277.3:c.209C>T

Likely Pathogenic

Met criteria codes **4**

PM2

PM3

PP4_Moderate

PP3

Evidence Links **1**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.209C>T (p.Ser70Phe) variant in PAH was reported in a Spanish patient with Mild HPA. A defect in the synthesis or regeneration in the pathways of 6R-BH4 was ruled out by analyzing urinary pterin levels and by measuring the dihydropteridine reductase activity (PMID 27121329). This variant was detected in trans with pathogenic variant p.Ala300Ser. It is absent in population databases and was predicted deleterious using in silico data. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP4 moderate, and PP3.

Met criteria codes

PM2			Absent from 1000G, ESP, and gnomAD.
PM3			Detected in trans with pathogenic variant p.Ala300Ser. Segregation analysis was done (PMID 27121329).
PubMed:21307867 			
PP4_Moderate			Reported in a Spanish patient with Mild HPA-NT. A defect in the synthesis or regeneration in the pathways of 6R-BH4 was ruled out by analyzing urinary pterin levels as well as by measuring the dihydropteridine reductase activity (PMID 27121329).
PP3			Predicted deleterious in SIFT, PolyPhen2, Mutation Taster, REVEL=0.832

Curation History



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