

Variant: *NM_000277.2(PAH):c.1069T>G (p.Cys357Gly)*

Version: 1.0

CA229329 [↗](#)

102514 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 0977199f-62f0-46c1-b092-5206646c9ea4

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HGVS expressions

NM_000277.2(PAH):c.1069T>G (p.Cys357Gly)

NM_000277.2:c.1069T>G

NC_000012.12:g.102843776A>C

CM000674.2:g.102843776A>C

NC_000012.11:g.103237554A>C

CM000674.1:g.103237554A>C

NC_000012.10:g.101761684A>C

NG_008690.1:g.78827T>G

NG_008690.2:g.119635T>G

ENST00000553106.6:c.1069T>G

ENST00000307000.7:c.1054T>G

ENST00000549247.6:n.828T>G

ENST00000551114.2:n.731T>G

ENST00000553106.5:c.1069T>G

ENST00000635477.1:c.173T>G

ENST00000635528.1:n.584T>G

NM_000277.1:c.1069T>G

NM_001354304.1:c.1069T>G

NM_000277.3:c.1069T>G

NM_001354304.2:c.1069T>G

Likely Pathogenic

Met criteria codes **4**

PP3

PP4

PM2

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.1069T>G (p.Cys357Gly) variant in PAH has been reported in 1 individual with classic PKU in trans with pathogenic variant p.R176X (PMID: 11139255). This variant is absent in population databases. Computational evidence supports a deleterious effect. In summary, this

variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4, PM2, PM3, PP3.

Met criteria codes

PP3			Predicted deleterious by PolyPhen, MutationTaster, SIFT.
PP4			Seen in an individual with classic PKU, in trans with R176X (pathogenic). PHE level >4 for all patients in study. Seen in an individual with classic PKU, in trans with R176X (pathogenic). PHE level >4 for all patients in study. PubMed:11139255 
PM2			Absent from controls.
PM3			in trans with R176X (pathogenic), paternal allele PMID: 11139255 Seen in an individual with classic PKU, in trans with R176X (pathogenic). PHE level >4 for all patients in study. PubMed:11139255 

Curation History

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