

Variant: *NM_000277.3:c.1129T>G*

Version: 1.0

CA16020946 [↗](#)

932278 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 39507c47-d45b-462f-9aaf-5205e361c94e

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HGVS expressions

NM_000277.3:c.1129T>G

NC_000012.12:g.102843716A>C

CM000674.2:g.102843716A>C

NC_000012.11:g.103237494A>C

CM000674.1:g.103237494A>C

NC_000012.10:g.101761624A>C

NG_008690.1:g.78887T>G

NG_008690.2:g.119695T>G

ENST00000553106.6:c.1129T>G

ENST00000307000.7:c.1114T>G

ENST00000549247.6:n.888T>G

ENST00000551114.2:n.791T>G

ENST00000553106.5:c.1129T>G

ENST00000635477.1:c.233T>G

ENST00000635528.1:n.644T>G

NM_000277.1:c.1129T>G

NM_000277.2:c.1129T>G

NM_001354304.1:c.1129T>G

NM_001354304.2:c.1129T>G

Likely Pathogenic

Met criteria codes **4**

PP4_Moderate PP3 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.1129T>G (p.Tyr377Asp) variant in PAH has been reported in 1 Armenian individual with classic PKU (BH4 deficiency ruled out) (PP4_Moderate; PMID: 21890392). This variant is absent in population databases (PM2). This variant was detected in trans with c.1066-

11G>A (Pathogenic in ClinVar) (PM3, PMID: 21890392). This variant is prediction to be deleterious by SIFT, PolyPhen, and MutationTaster, and REVEL = 0.932 (PP3). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP3, PP4_Moderate.

Met criteria codes

PP4_Moderate			PMID: 21890392 - T377D detected in Armenian patient (case #9) through sequence analysis of PAH exons (classic PKU, serum Phe = 20-30mg/dl, BH4 deficiency ruled out via HPLC to measure pterin concentration)
<div>PubMed:21890392 </div>			
PP3			Deleterious effect predicted by SIFT, PolyPhen, and MutationTaster. REVEL = 0.932.
PM2			Variant absent from population databases
PM3			PMID: 21890392 - This patient (case #8) carried the previously described variant c.1066-11G>A (VarID 607, P, 10 submitters) which was inherited on the paternal allele. Y377D inherited on the maternal allele. = 1.0 point

Not Met criteria codes

PM5			c.1130A>G (p.Y377C), not assertion in ClinVar (ClinVar ID: 102534). Curation in progress by PAH VCEP, currently classified as Likely Pathogenic.
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Curation History

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