

Variant: *NM_000277.3(PAH):c.1109A>G (p.Glu370Gly)*

Version: 1.0

CA386493290 [↗](#)

625290 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 57097d52-23f4-4ff3-ad9a-85e0bf487efd

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Published on: 2020/3/27

HGVS expressions

NM_000277.3:c.1109A>G

NM_000277.3(PAH):c.1109A>G (p.Glu370Gly)

NC_000012.12:g.102843736T>C

CM000674.2:g.102843736T>C

NC_000012.11:g.103237514T>C

CM000674.1:g.103237514T>C

NC_000012.10:g.101761644T>C

NG_008690.1:g.78867A>G

NG_008690.2:g.119675A>G

ENST00000553106.6:c.1109A>G

ENST00000307000.7:c.1094A>G

ENST00000549247.6:n.868A>G

ENST00000551114.2:n.771A>G

ENST00000553106.5:c.1109A>G

ENST00000635477.1:c.213A>G

ENST00000635528.1:n.624A>G

NM_000277.1:c.1109A>G

NM_000277.2:c.1109A>G

NM_001354304.1:c.1109A>G

NM_001354304.2:c.1109A>G

Likely Pathogenic

Met criteria codes **3**

PP4_Moderate

PM2

PM3

Not Met criteria codes **1**

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.1109A>G (p.Glu370Gly) variant in PAH has been reported in a Uygur patient with moderate PKU (BH4 deficiency ruled out) (PP4_Moderate; PMID: 31355225). This variant was detected with p.Val230Ile (Likely pathogenic in ClinVar) (PM3; PMID: 31355225). The variant is absent from population databases (PM2). Computational evidence is conflicting. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP4_Moderate.

Met criteria codes

PP4_Moderate			PMID: 31355225 - PCR and Sanger sequencing detected E370G in Uygur patient (Patient 83) with moderate PKU (Phe = 360-1200umol/L), BH4 deficiency ruled out.
<div>PubMed:31355225 </div>			
PM2			Variant absent from population databases.
PM3			Patient 83 carried the previously described mutation p.V230I (VarID 102784, LP, 10 submitters), parents were tested - variants confirmed in trans - 1.0 points

Not Met criteria codes

PP3			Predicted deleterious by SIFT and MutationTaster, predicted benign by PolyPhen2, REVEL = 0.744.
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Curation History

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