

Variant: *NM_000277.3:c.1105C>G*

Version: 1.0

CA16020941 [↗](#)

1327564 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 6c5abef4-0425-4068-89b1-32c3730a4636

Approved on: 2020/1/20

Published on: 2021/9/6

HGVS expressions

NM_000277.3:c.1105C>G

NC_000012.12:g.102843740G>C

CM000674.2:g.102843740G>C

NC_000012.11:g.103237518G>C

CM000674.1:g.103237518G>C

NC_000012.10:g.101761648G>C

NG_008690.1:g.78863C>G

NG_008690.2:g.119671C>G

ENST00000553106.6:c.1105C>G

ENST00000307000.7:c.1090C>G

ENST00000549247.6:n.864C>G

ENST00000551114.2:n.767C>G

ENST00000553106.5:c.1105C>G

ENST00000635477.1:c.209C>G

ENST00000635528.1:n.620C>G

NM_000277.1:c.1105C>G

NM_000277.2:c.1105C>G

NM_001354304.1:c.1105C>G

NM_001354304.2:c.1105C>G

Likely Pathogenic

Met criteria codes **3**

PP4_Moderate

PM2

PM3

Not Met criteria codes **1**

PP3

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.1105C>G (p.Leu369Val) variant in PAH has been reported in 2 individuals with mild HPA (BH4 deficiency excluded). (PP4_Moderate; PMID: 18299955). This variant is absent in population databases (PM2). This variant was detected with pathogenic variants p.T380M and

p.V230I (Parental analysis not reported, PM3). Computational evidence is conflicting. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM3.

Met criteria codes			
PP4_Moderate			Detected in 2 patients with mild HPA. BH4 deficiency excluded. PMID: 18299955
PM2			Absent from controls in ExAC, gnomAD, 1000 Genomes, ESP
PM3			Detected with T380M (P, multiple submitters) and V230I (P/LP). Parental analysis not reported.
Not Met criteria codes			
PP3			Computational evidence is conflicting: deleterious in SIFT; benign in PolyPhen2, MutationTaster, REVEL=0.598.

Curation History

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