

Variant: *NM_000277.3(PAH):c.452A>G (p.Asp151Gly)*

Version: 1.0

CA229554 [↗](#)

102683 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 3f8cd12f-4f9e-4eb7-b464-64d5b77c6e35

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HGVS expressions

NM_000277.3(PAH):c.452A>G (p.Asp151Gly)

NM_000277.3:c.452A>G

NC_000012.12:g.102866653T>C

CM000674.2:g.102866653T>C

NC_000012.11:g.103260431T>C

CM000674.1:g.103260431T>C

NC_000012.10:g.101784561T>C

NG_008690.1:g.55950A>G

NG_008690.2:g.96758A>G

ENST00000553106.6:c.452A>G

ENST00000307000.7:c.437A>G

ENST00000549111.5:n.548A>G

ENST00000551988.5:n.530+10809A>G

ENST00000553106.5:c.452A>G

NM_000277.1:c.452A>G

NM_000277.2:c.452A>G

NM_001354304.1:c.452A>G

NM_001354304.2:c.452A>G

Likely Pathogenic

Met criteria codes **3**

PP3_Strong

PP4

PM2_Supporting

Not Met criteria codes **1**

PM5

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Phenylketonuria Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PAH Version 2.0.0

[↗](#) **Criteria Specification Approval History**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.452A>G (p.Asp151Gly) variant in PAH is reported in a Spanish patient with phenylketonuria (PMID: 8981952); the second allele/genotype was not reported. It is absent from gnomAD. Multiple lines of computational evidence support a deleterious effect (REVEL=0.98). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2_supporting, PP3_strong, PP4.

Met criteria codes

PP3_Strong			Predicted deleterious in REVEL=0.98, SIFT, PP-2, MutationTaster
PP4			Reported in a Spanish patient with PKU (serum phenylalanine levels at diagnosis were >600 umol/liter and after exclusion of a defect in tetrahydrobiopterine metabolism) PMID: 8981952
PM2_Supporting			Absent from gnomAD v4.1.0

Not Met criteria codes

PM5			D151H has no interpretation in ClinVar
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Curation History

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