

Variant: *NM_000277.3(PAH):c.1156T>G (p.Tyr386Asp)*

Version: 1.0

CA229361 [↗](#)

102537 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 428af0db-e5b5-4ac8-8ef3-d61055a45a3a

Approved on: 2022/6/24

Published on: 2022/6/28

HGVS expressions

NM_000277.3:c.1156T>G

NM_000277.3(PAH):c.1156T>G (p.Tyr386Asp)

NC_000012.12:g.102843689A>C

CM000674.2:g.102843689A>C

NC_000012.11:g.103237467A>C

CM000674.1:g.103237467A>C

NC_000012.10:g.101761597A>C

NG_008690.1:g.78914T>G

NG_008690.2:g.119722T>G

ENST00000553106.6:c.1156T>G

ENST00000307000.7:c.1141T>G

ENST00000549247.6:n.915T>G

ENST00000551114.2:n.818T>G

ENST00000553106.5:c.1156T>G

ENST00000635477.1:c.260T>G

ENST00000635528.1:n.671T>G

NM_000277.1:c.1156T>G

NM_000277.2:c.1156T>G

NM_001354304.1:c.1156T>G

NM_001354304.2:c.1156T>G

Likely Pathogenic

Met criteria codes **5**

PM3_Supporting

PP3

PP4

PM2

PM5

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.1156T>G (p.Tyr386Asp) variant in PAH has been reported in 1 female, Italian patient - type of PKU not specified, serum Phe = 822umol/L; BH4 responsiveness was not specified (PMID: 23430918; PP4). This variant has been detected with R158Q - reported as

pathogenic in ClinVar (VarID:587), 14 submitters, phase unknown - 0.5 points (PMID: 23430918; PM3_Supporting). Another missense change at the same amino is pathogenic (p.Tyr386Cys). This variant is absent from population databases (PM2) and is predicted deleterious by SIFT, PolyPhen2, MutationTaster, and REVEL = 0.977 (PP3). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM5, PP3, PP4, PM3_Supporting.

Met criteria codes

PM3_Supporting			PMID: 23430918 - Y386D detected with R158Q - reported as pathogenic in ClinVar (VarID:587), 14 submitters, phase unknown - 0.5 points
PP3			Predicted deleterious by SIFT, PolyPhen2, MutationTaster, and REVEL = 0.977.
PP4			PMID: 23430918 - Y386D reported in 1 female, Italian patient - type of PKU not specified, serum Phe = 822umol/L; BH4 responsiveness was not specified
PM2			Variant absent from population databases.
PM5			At the same codon as c.1157A>G (p.Tyr386Cys) pathogenic by ClinGen PAH VCEP

Curation History

▼

▼

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.