

Variant: NM_000277.2(PAH):c.110T>C (p.Leu37Pro)

CA357242 [↗](#)

225133 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 42dfc728-4452-4eae-ae00-32fd711ab8ac

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HGVS expressions

NM_000277.2(PAH):c.110T>C (p.Leu37Pro)

NM_000277.2:c.110T>C

NC_000012.12:g.102912849A>G

CM000674.2:g.102912849A>G

NC_000012.11:g.103306627A>G

CM000674.1:g.103306627A>G

NC_000012.10:g.101830757A>G

NG_008690.1:g.9754T>C

NG_008690.2:g.50562T>C

NM_000277.1:c.110T>C

NM_001354304.1:c.110T>C

NM_000277.3:c.110T>C

ENST00000307000.7:c.95T>C

ENST00000546844.1:c.110T>C

ENST00000548677.2:n.197T>C

ENST00000548928.1:n.32T>C

ENST00000549111.5:n.206T>C

ENST00000550978.6:n.94T>C

ENST00000551337.5:c.110T>C

ENST00000551988.5:n.199T>C

ENST00000553106.5:c.110T>C

ENST00000635500.1:n.78T>C

Likely Pathogenic

Met criteria codes **4**

PM3

PM2

PP3

PP4

Evidence Links **2**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.110T>C (p.Leu37Pro) variant in PAH is reported in 1 patient with mild PKU. Assessment of BH4 deficiencies not reported. It was detected with a known pathogenic variant, p.R408W. (PMID: 24350308) This variant is absent from ExAC, gnomAD, 1000G, and ESP. A deleterious effect is predicted in SIFT, Polyphen-2, and MutationTaster. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP4, PP3.

Met criteria codes

PM3			Detected with R408W PMID: 24350308
			Patient genotype: p.R408W(c.1222C>T)/p.L37P(c.110C>T) PubMed:24350308 
PM2			Absent from ExAC, gnomAD, 1000G, ESP
PP3			Deleterious effect predicted in SIFT, Polyphen-2, MutationTaster
PP4			L37P detected in 1 patient with mild PKU. Assessment of BH4 deficiencies not reported. PMID: 24350308
			L37P detected in 1 patient with mild PKU. PubMed:24350308 
			Article in Chinese. PubMed:24078561 

Curation History

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