

Variant: NM_001354304.2:c.1016C>T

Version: 1.0

CA386493446 [↗](#)

1810387 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 51ea6579-cf3b-43d0-89ad-226c776fe307

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HGVS expressions

NM_001354304.2:c.1016C>T
NC_000012.12:g.102844385G>A
CM000674.2:g.102844385G>A
NC_000012.11:g.103238163G>A
CM000674.1:g.103238163G>A
NC_000012.10:g.101762293G>A
NG_008690.1:g.78218C>T
NG_008690.2:g.119026C>T
ENST00000553106.6:c.1016C>T
ENST00000307000.7:c.1001C>T
ENST00000549247.6:n.775C>T
ENST00000551114.2:n.678C>T
ENST00000553106.5:c.1016C>T
ENST00000635477.1:c.120C>T
ENST00000635528.1:n.531C>T
NM_000277.1:c.1016C>T
NM_000277.2:c.1016C>T
NM_001354304.1:c.1016C>T
NM_000277.3:c.1016C>T

Likely Pathogenic

Met criteria codes 4

PP3 PP4 PM2 PM3

Not Met criteria codes 1

PM5

Evidence Links 0

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information !

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

Evidence submitted by expert panel

Phenylketonuria VCEP

This c.1016 C>T (p.Ser339Phe) variant was detected in a patient with mPKU in trans with the pathogenic variant p. Ex6-96A>G (PMID: 28982351). This variant was absent in population databases. Multiple lines of computational evidence support a deleterious effect. In

summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM3, PM2, PP3, PP4.

Met criteria codes

PP3			Predicted to be damaging (SIFT), possibly damaging (PolyPhen2), Disease causing (MutationTaster). REVEL=0.716
PP4			Variant was detected in a patient affected with mPKU. Variable sites in patient genes were aligned with the corresponding sites from the respective parents. This was seen in trans with a pathogenic variant listed in ClinVar. Those with BH4 cofactor deficiency were excluded PMID: 28982351
PM2			This variant is absent from population databases gnomAD and ExAC
PM3			This variant was detected in trans with the pathogenic variant p. Ex6-96A>G (evaluated by an expert panel and seen in multiple entries in ClinVar) in a patient with mPKU (PMID: 28982351). points=1.

Not Met criteria codes

PM5			No other variants at this amino acid site were documented in ClinVar.
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Curation History



 

 

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