

Variant: *NM_001354304.2:c.1078G>T*

Version: 1.0

CA16020935 [↗](#)

1693236 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 89ab15ff-9068-43e7-8228-fca13e803639

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HGVS expressions

NM_001354304.2:c.1078G>T
NC_000012.12:g.102843767C>A
CM000674.2:g.102843767C>A
NC_000012.11:g.103237545C>A
CM000674.1:g.103237545C>A
NC_000012.10:g.101761675C>A
NG_008690.1:g.78836G>T
NG_008690.2:g.119644G>T
ENST00000553106.6:c.1078G>T
ENST00000307000.7:c.1063G>T
ENST00000549247.6:n.837G>T
ENST00000551114.2:n.740G>T
ENST00000553106.5:c.1078G>T
ENST00000635477.1:c.182G>T
ENST00000635528.1:n.593G>T
NM_000277.1:c.1078G>T
NM_000277.2:c.1078G>T
NM_001354304.1:c.1078G>T
NM_000277.3:c.1078G>T

Pathogenic

Met criteria codes **3**

PP4 PM2 PVS1

Not Met criteria codes **1**

PM3

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.1078G>T (p.Glu360Ter) nonsense variant occurs in exon 11 of 13 and is predicted to result in NMD. It absent from population databases, including gnomAD. It has been reported in at least one HPA patient (PMID: 10693064). In summary, this variant meets criteria

to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PM2, PP4.

Met criteria codes

PP4			One HPA patient was reported in PMID: 10693064 with Phe >180umol/L
PM2			The variant is absent from population databases including gnomAD, ExAC, 1000 Genomes, and ESP.
PVS1			The .1078G>T (p.Glu360Ter) nonsense variant occurs in exon 11 of 13 and is predicted to result in NMD.

Not Met criteria codes

PM3			E360X was identified in one allele of a patient in PMID: 10693064, however the other allele was not specified.
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Curation History

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