

Variant: NM_001354304.2:c.1204T>G

Version: 2.0

CA16020969 [↗](#)

987756 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8b454f5c-1d87-40ac-a8b5-c62fe71d8c7b

Approved on: 2024/11/17

Published on: 2024/11/18

HGVS expressions

NM_001354304.2:c.1204T>G
NC_000012.12:g.102840511A>C
CM000674.2:g.102840511A>C
NC_000012.11:g.103234289A>C
CM000674.1:g.103234289A>C
NC_000012.10:g.101758419A>C
NG_008690.1:g.82092T>G
NG_008690.2:g.122900T>G
ENST00000553106.6:c.1204T>G
ENST00000307000.7:c.1189T>G
ENST00000551114.2:n.866T>G
ENST00000553106.5:c.1204T>G
ENST00000635477.1:c.308T>G
ENST00000635528.1:n.719T>G
NM_000277.1:c.1204T>G
NM_000277.2:c.1204T>G
NM_001354304.1:c.1204T>G
NM_000277.3:c.1204T>G

Likely Pathogenic

Met criteria codes **3**

PM5_Supporting **PP3_Strong**
PM2_Supporting

Not Met criteria codes **2**

PP4 **PM3**

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.1204T>G (p.Phe402Val) variant in PAH is currently unreported in the literature. This variant is absent from population databases (PM2_Supporting), and is predicted deleterious by SIFT, PolyPhen2, MutationTester, and REVEL = 0.973 (PP3_Strong). Other missense variants [c.1204T>C (p.Phe402Leu); c.1205T>G (p.Phe402Cys)] in the same codon have been classified as likely pathogenic by the ClinGen Phenylketonuria Variant Curation Expert Panel (PM5_Supporting). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2_Supporting, PP3_Strong, PM5_supporting.

Met criteria codes

PM5_Supporting			At same codon as c.1204T>C (p.F402L), and c.1205T>G (p.F402C) - now LP by ClinGen PAH VCEP.
PP3_Strong			Predicted deleterious by SIFT, PolyPhen2, MutationTester, and REVEL = 0.973.
PM2_Supporting			Variant absent from gnomAD v4.1.0

Not Met criteria codes

PP4			This variant is currently unreported in the literature.
PM3			This variant is currently unreported in the literature.

Curation History

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