

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

Version: 1.0

CA16020968 [↗](#)

1327562 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8afe5778-b49b-4919-813c-e0af719d3ac3

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HGVS expressions

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

Uncertain Significance

Met criteria codes **3**

PP4_Moderate PP3 PM2

Not Met criteria codes **2**

PM3 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.1204T>A (p.Phe402Ile) variant in PAH has been detected in 1 Japanese patient with mild HPA, BH4 deficiency excluded (PMID: 21307867). This variant is absent from population databases (PM2), and is predicted deleterious by SIFT, PolyPhen2, MutationTaster, and

REVEL = 0.946 (PP3). In summary, this variant meets criteria to be classified as uncertain significance for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PP3, PP4_M.

Met criteria codes			
PP4_Moderate			F402I detected in 1 Japanese patient with mild HPA, serum Phe = 180-600umol/l - exact Phe level for patient not specified; included analysis of dihydropteridine reductase activity in red blood cells, biopterin loading test and/or pteridine analysis in urine. PMID: 21307867
PP3			Predicted deleterious by SIFT, PolyPhen2, MutationTaster, and REVEL = 0.946.
PM2			Variant absent from population databases.
Not Met criteria codes			
PM3			Genotype reported as "F402I/sm" where sm= severe mutation
PM5			At same codon as c.1204T>C (p.F402L), c.1204T>G (p.F402V), and c.1205T>G (p.F402V) - all curated as VUS by ClinGen PAH VCEP.

Curation History

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