

Variant: NM_001354304.2:c.920G>A

Version: 1.0

CA16020898 [↗](#)

1693235 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: e2f311ba-7dbb-425f-baa6-56501a3faf9c

Approved on: 2022/6/12

Published on: 2022/6/12

HGVS expressions

NM_001354304.2:c.920G>A

NC_000012.12:g.102846944C>T

CM000674.2:g.102846944C>T

NC_000012.11:g.103240722C>T

CM000674.1:g.103240722C>T

NC_000012.10:g.101764852C>T

NG_008690.1:g.75659G>A

NG_008690.2:g.116467G>A

ENST00000553106.6:c.920G>A

ENST00000307000.7:c.905G>A

ENST00000549247.6:n.679G>A

ENST00000551114.2:n.582G>A

ENST00000553106.5:c.920G>A

ENST00000635477.1:c.74-2513G>A

ENST00000635528.1:n.435G>A

NM_000277.1:c.920G>A

NM_000277.2:c.920G>A

NM_001354304.1:c.920G>A

NM_000277.3:c.920G>A

Likely Pathogenic

Met criteria codes **4**

PP4_Moderate

PP3

PM2

PM3_Strong

Evidence Links **2**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The NM_000277.3:c.920G>A (p.Gly307Asp) variant is a missense variant in exon 9/13 of PAH. The variant has been reported in confirmed trans with the p.R111* variant (Pathogenic per ClinGen PAH VCEP) in a Chinese patient with mild PKU (plasma Phe 10-20 mg/dl); BH4 deficiency was excluded by analysis of urinary pterins and dihydropteridine reductase activity in erythrocytes (PMID: 26322415)

(PP4_Moderate). It has also been found in presumed trans in 2 Polish cases: one patient with genotype F55Lfs/G307D with mild PKU and one patient with genotype R408W/G307D with unspecified Phe levels (PMID: 24350308) (PM3_Strong, 2 points total). The variant is absent from ethnically diverse control databases, including gnomAD/ExAC, 1000 Genomes, and ESP (PM2). The variant is predicted damaging by multiple in-silico missense predictors, including REVEL (REVEL score 0.983) (PP3). Classification: Likely Pathogenic Supporting Criteria: PM3; PM2; PP4_Moderate; PP3

Met criteria codes

PP4_Moderate			BH4 defects excluded in all patients. Single patient in this paper c.920G>A/C.331C>T (pathogenic) with mild PKU
			BH4 defects excluded in all patients. Single patient in this paper c.920G>A/C.331C>T (pathogenic) with mild PKU PubMed:26322415 
PP3			All computational evidence suggests damaging variant. REVEL=0.983
PM2			Not found in any population database
PM3_Strong			one patient F55Lfs/G307D with mild PKU and one patient R408W/G307D with unspecified Phe levels (PMID: 24350308). Also found in one patient with mild PKU and genotype R111*/G307D (PMID: 26322415)
			2 patients with c.920G>A in trans with pathogenic variants PubMed:24350308 

Curation History

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