

Variant: NM_000277.2(PAH):c.1024delG (p.Ala342Hisfs)

CA229279 [↗](#)

102475 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 4cc2d322-d860-4209-b3df-63e58bac3b07

Approved on: 2018-08-10

Published on: 2018-12-10

HGVS expressions

NM_000277.2(PAH):c.1024delG (p.Ala342Hisfs)

NM_000277.2:c.1024delG

NC_000012.12:g.102844378del

CM000674.2:g.102844378del

NC_000012.11:g.103238156del

CM000674.1:g.103238156del

NC_000012.10:g.101762286del

NG_008690.1:g.78226del

NG_008690.2:g.119034del

NM_000277.1:c.1024del

NM_000277.2:c.1024del

NM_001354304.1:c.1024del

NM_000277.3:c.1024del

ENST00000307000.7:c.1009del

ENST00000549247.6:n.783del

ENST00000551114.2:n.686del

ENST00000553106.5:c.1024del

ENST00000635477.1:n.128del

ENST00000635528.1:n.539del

Pathogenic

Met criteria codes **3**

PVS1

PP4_Moderate

PM2

Evidence Links **1**

Expert Panel

[Phenylketonuria VCEP](#) [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

PAH-specific ACMG/AMP criteria applied: PM2: Absent from ExAC, gnomAD, 1000G, ESP; PVS1: Frameshift variant; PP4_Moderate: Reported in patients with PAH deficiency. Bh4 defects excluded. (PMID:9634518). In summary this variant meets criteria to be classified as pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PM2, PVS1, PP4_Moderate).

Met criteria codes

PVS1			Frameshift variant
PP4_Moderate			Reported in patients with PAH deficiency. Bh4 defects excluded. <div>A342fsdelG detected. PAH deficiency had been assessed after exclusion of a defect in tetrahydrobiopterin metabolism. PubMed:9634518 </div>
PM2			Absent from ExAC, gnomAD, 1000G, ESP

Curation History

  

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification 	Condition	Published Date	Version 	Criteria Specification	Gene
View	NM_000277.2(PAH):c.1024delG (p.Ala3...	Pathogenic	Phenylketonuria 	2019-08-17	1.0	-	PAH 

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.