

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

Version: 1.0

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

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HGVS expressions

NM_000277.2:c.1024delG

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

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

ENST00000553106.6:c.1024del

ENST00000307000.7:c.1009del

ENST00000549247.6:n.783del

ENST00000551114.2:n.686del

ENST00000553106.5:c.1024del

ENST00000635477.1:c.128del

ENST00000635528.1:n.539del

NM_000277.1:c.1024del

NM_000277.2:c.1024del

NM_001354304.1:c.1024del

NM_000277.3:c.1024del

NM_001354304.2:c.1024del

Pathogenic

Met criteria codes **3**

PM2

PVS1

PP4_Moderate

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

PM2	✓	Absent from ExAC, gnomAD, 1000G, ESP
PVS1	✓	Frameshift variant
PP4_Moderate	✓	Reported in patients with PAH deficiency. Bh4 defects excluded.

A342fsdelG detected. PAH deficiency had been assessed after exclusion of a defect in tetrahydrobiopterin metabolism.

[PubMed:9634518](#) 

Curation History

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