

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** **PP4\_Moderate** **PVS1**

**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

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

#### Curation History [↗](#)

Showing 1 to 1 of 1 rows

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