

Variant: *NM\_000277.2(PAH):c.806delT (p.Ile269Thrfs)*

Version: 1.0

CA229778 [↗](#)

102844 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal recessive inheritance

**UUID:** f5d8dc3f-dba0-4dc9-98cd-4a22b6a11a83

**Approved on:** 2018-08-10

**Published on:** 2019-08-17

### *HGVS expressions*

**NM\_000277.2:c.806delT**

NM\_000277.2(PAH):c.806delT (p.Ile269Thrfs)

NC\_000012.12:g.102852851del

CM000674.2:g.102852851del

NC\_000012.11:g.103246629del

CM000674.1:g.103246629del

NC\_000012.10:g.101770759del

NG\_008690.1:g.69752del

NG\_008690.2:g.110560del

ENST00000553106.6:c.806del

ENST00000307000.7:c.791del

ENST00000549247.6:n.565del

ENST00000553106.5:c.806del

NM\_000277.1:c.806del

NM\_000277.2:c.806del

NM\_001354304.1:c.806del

NM\_000277.3:c.806del

NM\_001354304.2:c.806del

**Pathogenic**

Met criteria codes **3**

**PVS1** **PP4** **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: PVS1: Frameshift variant; PM2: Extremely low frequency. gnomAD MAF=0.00007.; PP4: Detected in a PKU patient. BH4 deficiency not assessed. (PMID:9012412). 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: (PVS1, PM2, PP4).**

### Met criteria codes

<b>PVS1</b>	✓	Frameshift variant
<b>PP4</b>	✓	Detected in a PKU patient. BH4 deficiency not assessed. <hr/> I269fsdelT detected on 1 PKU chromosome. <a href="#">PubMed:9012412</a>
<b>PM2</b>	✓	Extremely low frequency. gnomAD MAF=0.00007.

### Curation History [↗](#)



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See Report	Preferred Variant Title	Classification <sup>i</sup>	Condition	Published Date	Version <sup>i</sup>	Criteria Specification	Gene
<a href="#">View</a>	NM_000277.2(PAH):c.806delT (p.Ile269...)	<b>Pathogenic</b>	<a href="#">Phenylketonuria</a>	2019-08-17	1.0	-	<a href="#">PAH</a>

Showing 1 to 1 of 1 rows

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