

Variant: *NM\_000277.2(PAH):c.355C>T (p.Pro119Ser)*

Version: 1.0

CA220582 [↗](#)

92741 (ClinVar) [↗](#)

**Gene:** PAH (HGNC:5053)

**Condition:** phenylketonuria (MONDO:0009861)

**Inheritance Mode:** Autosomal recessive inheritance

**UUID:** 38a183ee-7e53-4c24-a250-50958f390c78

**Approved on:** 2018-07-29

**Published on:** 2019-04-06

### *HGVS expressions*

**NM\_000277.2:c.355C>T**

NM\_000277.2(PAH):c.355C>T (p.Pro119Ser)

NC\_000012.12:g.102877548G>A

CM000674.2:g.102877548G>A

NC\_000012.11:g.103271326G>A

CM000674.1:g.103271326G>A

NC\_000012.10:g.101795456G>A

NG\_008690.1:g.45055C>T

NG\_008690.2:g.85863C>T

NM\_000277.1:c.355C>T

NM\_001354304.1:c.355C>T

NM\_000277.3:c.355C>T

ENST00000307000.7:c.340C>T

ENST00000549111.5:n.451C>T

ENST00000550978.6:n.339C>T

ENST00000551337.5:c.355C>T

ENST00000551988.5:n.444C>T

ENST00000553106.5:c.355C>T

**Likely Pathogenic**

Met criteria codes **3**

PP3 PP4\_Moderate PM3\_Strong

Not Met criteria codes **1**

PM2

Evidence Links **2**

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: PP3:** in silico analysis supportive of damaging effect; **PM3\_Strong:** In trans with R261Q (PMID 21147011), and in trans with IVS2+1G>A (PMID 12655554) (PMID:21147011; PMID:12655554); **PP4\_Moderate:** BH4 deficiency excluded

(PMID:21147011). In summary this variant meets criteria to be classified as likely pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PP3, PM3\_Strong, PP4\_Moderate).

#### Met criteria codes

<b>PP3</b>	✓	in silico analysis supportive of damaging effect
<b>PP4_Moderate</b>	✓	BH4 deficiency excluded BH4 defect excluded at NBS and by sequencing of BH4 associated genes <a href="#">PubMed:21147011</a>
<b>PM3_Strong</b>	✓	In trans with R261Q (PMID 21147011), and in trans with IVS2+1G>A (PMID 12655554) in trans with IVS2+1G>A <a href="#">PubMed:12655554</a> in trans with R261Q <a href="#">PubMed:21147011</a>

#### Not Met criteria codes

<b>PM2</b>	✗	PAH specific specifications state PM2 criteria as 0.02% (AF=0.0002)
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#### Curation History [↗](#)

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