

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

UID: 38a183ee-7e53-4c24-a250-50958f390c78

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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**

PM3_Strong

PP4_Moderate

PP3

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

PM3_Strong ✓ In trans with R261Q (PMID 21147011), and in trans with IVS2+1G>A (PMID 12655554)

in trans with IVS2+1G>A [PubMed:12655554](#)

in trans with R261Q [PubMed:21147011](#)

PP4_Moderate ✓ BH4 deficiency excluded

BH4 defect excluded at NBS and by sequencing of BH4 associated genes [PubMed:21147011](#)

PP3 ✓ in silico analysis supportive of damaging effect

Not Met criteria codes

PM2 ✗ PAH specific specifications state PM2 criteria as 0.02% (AF=0.0002)

Curation History [↗](#)

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