

Variant: *NM_000277.1(PAH):c.609C>G (p.Cys203Trp)*

Version: 1.0

CA229647 [↗](#)

102755 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 0aa6ba38-c16b-4adc-86a5-616bdef1424f

Approved on: 2020/2/22

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HGVS expressions

NM_000277.1:c.609C>G

NM_000277.1(PAH):c.609C>G (p.Cys203Trp)

NC_000012.12:g.102855233G>C

CM000674.2:g.102855233G>C

NC_000012.11:g.103249011G>C

CM000674.1:g.103249011G>C

NC_000012.10:g.101773141G>C

NG_008690.1:g.67370C>G

NG_008690.2:g.108178C>G

ENST00000553106.6:c.609C>G

ENST00000307000.7:c.594C>G

ENST00000549111.5:n.705C>G

ENST00000553106.5:c.609C>G

NM_000277.2:c.609C>G

NM_001354304.1:c.609C>G

NM_000277.3:c.609C>G

NM_001354304.2:c.609C>G

Likely Pathogenic

Met criteria codes 4

PM3_Supporting

PP4_Moderate

PP3

PM2

Not Met criteria codes 1

PM5

Evidence Links 0

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.609C>G (p.Cys203Trp) variant in PAH has been reported in 1 individual with PKU (BH4 deficiency excluded) detected with pathogenic variant c.1315+1G>A (P 13 submitters) parental analysis not performed. (PMID: 23430918). This variant has extremely low frequency in

1000G (MAF=0.0004) and is absent from gnomAD. Computational evidence supports a deleterious effect. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM3_supporting, PP3.

Met criteria codes

PM3_Supporting			Detected with c.1315+1G>A (P 13 submitters). parental analysis not performed
PP4_Moderate			detected in a patient with PKU. primary BH4 deficiency excluded. PMID: 23430918
PP3			Predicted deleterious in SIFT, PolyPhen2, MutationTaster, REVEL=0.91
PM2			MAF=0.0004 in 1000G, absent in ExAC, gnomAD, ESP

Not Met criteria codes

PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History

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