Variant: NM_000277.3(PAH):c.479A>C (p.Gln160Pro)

Version: 1.0

CA229573

102695 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 8a149ff5-cf7d-4456-928b-27aeece7ef71
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HGVS expressions

NM_000277.3:c.479A>C
NM_000277.3(PAH):c.479A>C (p.Gln160Pro)
NC_000012.12:g.102866626T>G
CM000674.2:g.102866626T>G
NC_000012.11:g.103260404T>G
CM000674.1:g.103260404T>G
NC_000012.10:g.101784534T>G
NG_008690.1:g.55977A>C
NG_008690.2:g.96785A>C
ENST00000553106.6:c.479A>C
ENST00000307000.7:c.464A>C
ENST00000549111.5:n.575A>C
ENST00000551988.5:n.530+10836A>C
ENST00000553106.5:c.479A>C
NM_000277.1:c.479A>C
NM_000277.2:c.479A>C
NM_001354304.1:c.479A>C
NM_001354304.2:c.479A>C

Uncertain Significance

Met criteria codes 3

PM2_Supporting PP3 PP4

Not Met criteria codes 2

PM3 PM5

Evidence Links 0

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Phenylketonuria Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PAH Version 2.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.479A>C (p.Gln160Pro) variant in PAH is reported in a German patient with PAH deficiency (PMID: 12655553); the second allele/genotype was not reported. It is absent from gnomAD. Computational evidence supports a deleterious effect (REVEL=0.668). In summary, this variant meets criteria to be classified as uncertain significance for PAH. PAH-specific ACMG/AMP criteria applied: PM2_supporting, PP3, PP4.

Met criteria codes

PM2_Supporting	✔	Absent from gnomAD v4.1.0
PP3	✔	REVEL=0.668
PP4	✔	Reported in a German patient with PAH deficiency, BH4 deficiency not reportedly ruled out. PMID: 12655553

Not Met criteria codes

PM3	✖	2nd allele not reported
PM5	✖	only variant found in this codon in ClinVar.

Curation History

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