Variant: NM_000277.2(PAH):c.464G>C (p.Arg155Pro)

Version: 1.0

CA229561

102687 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: a532eae1-862f-4d90-b69e-5e590a9f7512
Approved on: 2018-08-10
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HGVS expressions

NM_000277.2:c.464G>C
NM_000277.2(PAH):c.464G>C (p.Arg155Pro)
NC_000012.12:g.102866641C>G
CM000674.2:g.102866641C>G
NC_000012.11:g.103260419C>G
CM000674.1:g.103260419C>G
NC_000012.10:g.101784549C>G
NG_008690.1:g.55962G>C
NG_008690.2:g.96770G>C
NM_000277.1:c.464G>C
NM_001354304.1:c.464G>C
NM_000277.3:c.464G>C
ENST00000307000.7:c.449G>C
ENST00000549111.5:n.560G>C
ENST00000551988.5:n.530+10821G>C
ENST00000553106.5:c.464G>C

Likely Pathogenic

Met criteria codes 4

PP3 **PM3** **PM2** **PP4_Moderate**

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: PM2: absent from ExAC, gnomAD, 1000G, ESP. PAGE MAF=0.00066; PP3: Deleterious effect predicted in SIFT, Polyphen-2, MutationTaster. REVEL=0.967; PP4_Moderate: Detected in a patient with classic PKU. Cofactor deficiency excluded. (PMID:10679941); PM3: Detected in trans with R408W (P) (PMID:10679941). 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: (PM2, PP3, PP4_Moderate, PM3).

Met criteria codes

PP3	✓	Deleterious effect predicted in SIFT, Polyphen-2, MutationTaster. REVEL=0.967
PM3	✓	Detected in trans with R408W (P) Detected in trans with R408W PubMed:10679941
PM2	✓	absent from ExAC, gnomAD, 1000G, ESP. PAGE MAF=0.00066
PP4_Moderate	✓	Detected in a patient with classic PKU. Cofactor deficiency excluded. 302 PKU or HPA patients in 290 families were analyzed. Most of the patients were identified by neonatal screening. Cofactor deficiency was excluded by the BH4 test. Detected in a patient (SD) with classic PKU. PubMed:10679941

Curation History

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