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Variant: NM_001354304.2:c.679C>G

Version: 1.0

CA386296582

1065379 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 0c78dc36-0ebe-4bea-9d50-6ff0856bbb8f
Approved on: 2020/12/9
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HGVS expressions

NM_001354304.2:c.679C>G
NC_000012.12:g.102855163G>C
CM000674.2:g.102855163G>C
NC_000012.11:g.103248941G>C
CM000674.1:g.103248941G>C
NC_000012.10:g.101773071G>C
NG_008690.1:g.67440C>G
NG_008690.2:g.108248C>G
ENST00000553106.6:c.679C>G
ENST00000307000.7:c.664C>G
ENST00000549111.5:n.775C>G
ENST00000553106.5:c.679C>G
NM_000277.1:c.679C>G
NM_000277.2:c.679C>G
NM_001354304.1:c.679C>G
NM_000277.3:c.679C>G

Likely Pathogenic

Met criteria codes 4

PM3 PM2 PP3 PP4

Evidence Links 0

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

This c.679C>G (p.Leu227Val) variant in PAH was reported in trans with pathogenic variant p.Arg243Gln in 1 patient with PAH deficiency (1134µmol/L Phe) (PMID: 28982351). Computational evidence for this missense variant is predicted to be damaging (SIFT), probably damaging (PolyPhen2), and disease causing (MutationTaster). This variant is absent from population databases gnomAD, 1000 Genomes, and ESP. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PP3, PM3, PP4.

Met criteria codes

PM3	✔	This variant was detected in trans with the pathogenic PAH variant p.Arg243Gln (1.0pt) PMID: 28982351
PM2	✔	This variant is absent from population databases gnomAD,1000 Genomes, and ESP
PP3	✔	Computational evidence for this missense variant is predicted to be damaging (SIFT), probably damaging (PolyPhen2), and disease causing (MutationTaster).
PP4	✔	This variant was documented 1 time in a patient with PAH deficiency (1134µmol/L Phe) in the Han Chinese population. Patients with BH4 cofactor deficiency were excluded by BH4 loading. PMID: 28982351

Curation History

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