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Variant: NM_001354304.2:c.563G>T

Version: 1.0

CA386296833

1065370 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 93378ff3-a0f5-476d-9386-3bbd7a51e0f0
Approved on: 2020/11/16
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HGVS expressions

NM_001354304.2:c.563G>T
NC_000012.12:g.102855279C>A
CM000674.2:g.102855279C>A
NC_000012.11:g.103249057C>A
CM000674.1:g.103249057C>A
NC_000012.10:g.101773187C>A
NG_008690.1:g.67324G>T
NG_008690.2:g.108132G>T
ENST00000553106.6:c.563G>T
ENST00000307000.7:c.548G>T
ENST00000549111.5:n.659G>T
ENST00000551988.5:n.584G>T
ENST00000553106.5:c.563G>T
NM_000277.1:c.563G>T
NM_000277.2:c.563G>T
NM_001354304.1:c.563G>T
NM_000277.3:c.563G>T

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.563G>T (p.Gly188Val) variant in PAH was reported in trans with pathogenic variant p.Val399= in 1 patient with PAH deficiency (>120 µmol/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, PM3, PP3, PP4.

Met criteria codes

PM3	✔	Detected in trans with p.Val399= (P) 1.0pts PMID: 28982351
PM2	✔	Absent from population databases gnomAD, 1000 Genomes and ESP.
PP3	✔	Damaging in SIFT, PolyPhen2 and MutationTaster
PP4	✔	Detected in a patient with PAH deficiency (>120 µmol/L Phe). Patients with BH4 cofactor deficiency were excluded by BH4 loading. PMID: 28982351

Curation History

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