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Variant: NM_001354304.2:c.311C>T

Version: 1.0

CA386304006

1810384 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: bdfd24a5-2b03-45b5-9fee-9cc87e5acd76
Approved on: 2022/7/30
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HGVS expressions

NM_001354304.2:c.311C>T
NC_000012.12:g.102894776G>A
CM000674.2:g.102894776G>A
NC_000012.11:g.103288554G>A
CM000674.1:g.103288554G>A
NC_000012.10:g.101812684G>A
NG_008690.1:g.27827C>T
NG_008690.2:g.68635C>T
ENST00000553106.6:c.311C>T
ENST00000307000.7:c.296C>T
ENST00000546844.1:c.311C>T
ENST00000548928.1:n.233C>T
ENST00000549111.5:n.407C>T
ENST00000550978.6:c.295C>T
ENST00000551337.5:c.311C>T
ENST00000551988.5:n.400C>T
ENST00000553106.5:c.311C>T
NM_000277.1:c.311C>T
NM_000277.2:c.311C>T
NM_001354304.1:c.311C>T
NM_000277.3:c.311C>T

Likely Pathogenic

Met criteria codes 3
PM5 PM2 PP4_Moderate

Not Met criteria codes 1
PP3

Evidence Links 0

Expert Panel
Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.311C>T (p.Ala104Val) missense variant in PAH has been reported in 1 Chinese patient with classic PKU (Phe >1200umol/L; BH4 deficiency excluded) (PMID: 29499199; PP4_Moderate). This variant is absent from population databases (PM2), and is located at same location a c.311C>A (p.Ala104Asp), reported as Pathogenic in ClinVar (VarID:102650). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM5.

Met criteria codes

PM5	✔	At same location as NM_000277.3(PAH):c.311C>A (p.Ala104Asp); reported as Pathogenic in ClinVar (VarID:102650)
PM2	✔	Variant absent from population databases.
PP4_Moderate	✔	PMID: 29499199 - A104V detected in 1 Chinese patient with CPKU (serum Phe >1200umol/L); BH4 deficiency excluded via urinary pterin analysis with HPLC

Not Met criteria codes

PP3	✖	Computational evidence is conflicting, predicted benign by by SIFT and PolyPhen2, predicted deleterious by MutationTaster, REVEL = 0.508

Curation History

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