Variant: NM_000277.2(PAH):c.1242C>T (p.Tyr414=)

Version: 1.0

CA200893

102577 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 349736d7-05a8-4ec0-9b30-04fa24dd5529
Approved on: 2018-08-10
Published on: 2019-04-06

HGVS expressions

NM_000277.2:c.1242C>T
NM_000277.2(PAH):c.1242C>T (p.Tyr414=)
NC_000012.12:g.102840473G>A
CM000674.2:g.102840473G>A
NC_000012.11:g.103234251G>A
CM000674.1:g.103234251G>A
NC_000012.10:g.101758381G>A
NG_008690.1:g.82130C>T
NG_008690.2:g.122938C>T
NM_000277.1:c.1242C>T
NM_001354304.1:c.1242C>T
NM_000277.3:c.1242C>T
ENST00000307000.7:c.1227C>T
ENST00000551114.2:n.904C>T
ENST00000553106.5:c.1242C>T
ENST00000635477.1:n.346C>T
ENST00000635528.1:n.757C>T

Benign

Met criteria codes 2

BS2 BS1

Not Met criteria codes 1

BP4

Evidence Links 0

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: BS1: MAF=0.01361 in ENF from gnomAD; BS2: 19 homozygotes in gnomAD. In summary this variant meets criteria to be classified as benign for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (BS1, BS2).

Met criteria codes

BS2	✔	19 homozygotes in gnomAD
BS1	✔	MAF=0.01361 in ENF from gnomAD

Not Met criteria codes

BP4	✖	Tolerated and benign in SIFT, Polyphen2. Disease causing in MutationTaster.

Curation History

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