Variant: NM_000277.1:c.772C>T

Version: 1.0

CA6748843

590340 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 254ed141-a219-404a-8c94-1fd2aceb8103
Approved on: 2018-08-13
Published on: 2019-04-06

HGVS expressions

NM_000277.1:c.772C>T
NC_000012.12:g.102852885G>A
CM000674.2:g.102852885G>A
NC_000012.11:g.103246663G>A
CM000674.1:g.103246663G>A
NC_000012.10:g.101770793G>A
NG_008690.1:g.69718C>T
NG_008690.2:g.110526C>T
NM_000277.2:c.772C>T
NM_001354304.1:c.772C>T
NM_000277.3:c.772C>T
ENST00000307000.7:c.757C>T
ENST00000549247.6:n.531C>T
ENST00000553106.5:c.772C>T

Likely Benign

Met criteria codes 2

BS1 BP7

Not Met criteria codes 1

PM2

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: > PAH specific guidelines of AF-0.0002 (0.02%); BP7: No deleterious effect predicted.. In summary this variant meets criteria to be classified as likely benign for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (BS1, BP7).

Met criteria codes

BS1	✔	> PAH specific guidelines of AF-0.0002 (0.02%)

BP7	✔	No deleterious effect predicted.

Not Met criteria codes

PM2	✖	> PAH specific guidelines of AF-0.0002 (0.02%)

Curation History

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