

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

CA6748843 [↗](#)

590340 (ClinVar) [↗](#)

Gene: PAH ([HGNC:5053](#))

Condition: phenylketonuria ([MONDO:0009861](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 254ed141-a219-404a-8c94-1fd2aceb8103

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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**

BP7 **BS1**

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

BP7			No deleterious effect predicted.
BS1			> PAH specific guidelines of AF-0.0002 (0.02%)
Not Met criteria codes			
PM2			> PAH specific guidelines of AF-0.0002 (0.02%)

Curation History 			
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