

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

UID: 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 [↗](#)



Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.