

Variant: NM_000277.2(PAH):c.963C>T (p.Leu321=)

CA229873 [↗](#)

102911 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 31e57f08-7be8-46c7-902d-8429b753f57c

Approved on: 2018-07-29

Published on: 2018-12-10

HGVS expressions

NM_000277.2:c.963C>T

NM_000277.2(PAH):c.963C>T (p.Leu321=)

NC_000012.12:g.102846901G>A

CM000674.2:g.102846901G>A

NC_000012.11:g.103240679G>A

CM000674.1:g.103240679G>A

NC_000012.10:g.101764809G>A

NG_008690.1:g.75702C>T

NG_008690.2:g.116510C>T

NM_000277.1:c.963C>T

NM_001354304.1:c.963C>T

NM_000277.3:c.963C>T

ENST00000307000.7:c.948C>T

ENST00000549247.6:n.722C>T

ENST00000551114.2:n.625C>T

ENST00000553106.5:c.963C>T

ENST00000635477.1:n.74-2470C>T

ENST00000635528.1:n.478C>T

Benign

Met criteria codes **4**

BS2

BS1

BS3_Supporting

BP7

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: >0.02% as set by the PAH specific specifications; BP7: ; BS3_Supporting: cDNA method demonstrates 98% and intinic system demonstrates 81% residual enzyme activity; BS2: 38 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, BP7, BS3_Supporting, BS2).

Met criteria codes			
BS2			38 homozygotes in gnomAD
BS1			>0.02% as set by the PAH specific specifications
BS3_Supporting			cDNA method demonstrates 98% and intinic system demonstrates 81% residual enzyme activity
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 





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▼

See Report

◀▶

Preferred Variant Title

◀▶

Classification 

◀▶

Condition

◀▶

Published Date

◀▶

Version 

Criteria Specification

◀▶

Gene

No matching records found

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