The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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

Version: 1.0

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: 2019-04-06

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

BS3_Supporting BS2 BS1 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		
BS3_Supporting	✔	cDNA method demonstrates 98% and intinic system demonstrates 81% residual enzyme activity
BS2	✔	38 homozygotes in gnomAD
BS1	✔	>0.02% as set by the PAH specific specifications
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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