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.1278T>C (p.Asn426=)

Version: 1.0

CA145978

92732 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 66eeb668-a8a7-49cb-a6cd-26afcc800972
Approved on: 2018-08-10
Published on: 2019-04-06

HGVS expressions

NM_000277.2:c.1278T>C
NM_000277.2(PAH):c.1278T>C (p.Asn426=)
NC_000012.12:g.102840437A>G
CM000674.2:g.102840437A>G
NC_000012.11:g.103234215A>G
CM000674.1:g.103234215A>G
NC_000012.10:g.101758345A>G
NG_008690.1:g.82166T>C
NG_008690.2:g.122974T>C
NM_000277.1:c.1278T>C
NM_001354304.1:c.1278T>C
NM_000277.3:c.1278T>C
ENST00000307000.7:c.1263T>C
ENST00000551114.2:n.940T>C
ENST00000553106.5:c.1278T>C
ENST00000635477.1:n.382T>C
ENST00000635528.1:n.793T>C

Benign

Met criteria codes 2

BP4 BA1

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: BA1: MAF=0.16641; BP4: no impact on gene in SIFT, Polyphen2, MutationTaster. 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: (BA1, BP4).

Met criteria codes

BP4	✔	no impact on gene in SIFT, Polyphen2, MutationTaster.
BA1	✔	MAF=0.16641

Curation History

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