

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

UID: 66eeb668-a8a7-49cb-a6cd-26afcc800972

Approved on: 2018-08-10

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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**

BA1 BP4

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

BA1



MAF=0.16641

BP4



no impact on gene in SIFT, Polyphen2, MutationTaster.

Curation History [↗](#)

Showing 1 to 1 of 1 rows

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