

*Variant: NM\_000277.2(PAH):c.1278T>C (p.Asn426=)*

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:** 2018-12-10

### *HGVS expressions*

**NM\_000277.2(PAH):c.1278T>C (p.Asn426=)**

**NM\_000277.2:c.1278T>C**

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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