

Variant: *NM_000277.3(PAH):c.786C>T (p.Val262=)*

Version: 1.0

CA242471869 [↗](#)

883189 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 251ff915-9b59-4b03-8690-7ae55aea4c86

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HGVS expressions

NM_000277.3(PAH):c.786C>T (p.Val262=)

NM_000277.3:c.786C>T

NC_000012.12:g.102852871G>A

CM000674.2:g.102852871G>A

NC_000012.11:g.103246649G>A

CM000674.1:g.103246649G>A

NC_000012.10:g.101770779G>A

NG_008690.1:g.69732C>T

NG_008690.2:g.110540C>T

ENST00000553106.6:c.786C>T

ENST00000307000.7:c.771C>T

ENST00000549247.6:n.545C>T

ENST00000553106.5:c.786C>T

NM_000277.1:c.786C>T

NM_000277.2:c.786C>T

NM_001354304.1:c.786C>T

NM_001354304.2:c.786C>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **1**

BP7

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The NM_000277.3:c.786C>T in PAH is a synonymous (silent) variant (p.Val262=) that is not predicted to impact splicing. To our knowledge, this variant has not been reported in the literature and results of functional studies are unavailable. This variant is absent in gnomAD v2.1.1 (PM2_Supporting). There is a ClinVar entry for this variant (Variation ID: 883189, 1 star review status) with two submitters classifying the variant as a variant of uncertain significance and one submitter classifying the variant as likely benign. In summary, this

variant meets the criteria to be classified as a variant of uncertain significance for PAH deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen PAH Variant Curation Expert Panel (Specifications Version 2.0): PM2_Supporting.

Met criteria codes

PM2_Supporting ⓘ ✓ Absent in gnomad

Not Met criteria codes

BP7 ⓘ ✗ synonymous (silent) variant that is (p.Val262=) that is not predicted to impact splicing but nucleotide is conserved from humans through zebrafish

Curation History ↗



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