

Variant: NM_000277.2(PAH):c.735G>A (p.Val245=)

CA145982 [↗](#)

92748 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 9cca2153-6d64-4ab3-923e-39673d123cd3

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HGVS expressions

NM_000277.2:c.735G>A

NM_000277.2(PAH):c.735G>A (p.Val245=)

NC_000012.12:g.102852922C>T

CM000674.2:g.102852922C>T

NC_000012.11:g.103246700C>T

CM000674.1:g.103246700C>T

NC_000012.10:g.101770830C>T

NG_008690.1:g.69681G>A

NG_008690.2:g.110489G>A

NM_000277.1:c.735G>A

NM_001354304.1:c.735G>A

NM_000277.3:c.735G>A

ENST00000307000.7:c.720G>A

ENST00000549247.6:n.494G>A

ENST00000553106.5:c.735G>A

Benign

Met criteria codes **3**

BP4

BA1

BS2

Evidence Links **0**

Expert Panel

[Phenylketonuria VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.735G>A (p.Val245=) variant in PAH has a MAF of 0.29058 in ExAC (BA1; <http://exac.broadinstitute.org>) with 6,524 homozygotes (BS2). This is a synonymous variant, predicted tolerated and benign in SIFT, Polyphen. MutationTaster predicted polymorphism with no abrogation of splice sites (BP4). In summary, this variant meets criteria to be classified as benign.

Met criteria codes

BP4



			Synonymous variant, predicted tolerated and benign in SIFT, Polyphen. MutationTaster predicted polymorphism with no abrogation of splice sites
BA1			AF is 0.29058 in ExAC with 6,524 homozygotes. highest MAF is 0.76290 in EAS from 1000G
BS2			Reported in the homozygous state in large population databases

Curation History 		
		
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