

Variant: NM_000277.1(PAH):c.1162G>A (p.Val388Met)

CA251543 [↗](#)

619 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: c18b654a-1bd0-428b-b43f-2711b92f28a1

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HGVS expressions

NM_000277.1(PAH):c.1162G>A (p.Val388Met)

NM_000277.1:c.1162G>A

NC_000012.12:g.102843683C>T

CM000674.2:g.102843683C>T

NC_000012.11:g.103237461C>T

CM000674.1:g.103237461C>T

NC_000012.10:g.101761591C>T

NG_008690.1:g.78920G>A

NG_008690.2:g.119728G>A

NM_000277.2:c.1162G>A

NM_001354304.1:c.1162G>A

NM_000277.3:c.1162G>A

ENST00000307000.7:c.1147G>A

ENST00000549247.6:n.921G>A

ENST00000551114.2:n.824G>A

ENST00000553106.5:c.1162G>A

ENST00000635477.1:n.266G>A

ENST00000635528.1:n.677G>A

Pathogenic

Met criteria codes 5

PP3

PP4

PM2

PM3

PS3

Evidence Links 1

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: PM2: Extremely low in ExAC and 1000 genomes (PMID:9860305); PS3: PAH activity in COS cell expression system 15% (PMID:9860305); PM3: Compound het with severe mutation (PMID:9860305); PP3: ; PP4: Reported in patient with classic PKU (PMID:9860305). In summary this variant meets criteria to be classified as pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PM2, PS3, PM3, PP3, PP4).

Met criteria codes

PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			Reported in patient with classic PKU
Compound het with severe mutation, and classic pku PubMed:9860305 			
PM2			Extremely low in ExAC and 1000 genomes
1.2% Japanese PubMed:9860305 			
PM3			Compound het with severe mutation
Compound het with severe mutation, showed 8% activity, and classic pku PubMed:9860305 			
PS3			PAH activity in COS cell expression system 15%
PAH activity in COS cell expression system 15% PubMed:9860305 			

Curation History

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