

Variant: *NM_000277.2(PAH):c.386A>T (p.Asp129Val)*

Version: 1.2

CA229529 [↗](#)

102662 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 0f4335db-54e0-4e3f-9108-f651780b39f9

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HGVS expressions

NM_000277.2:c.386A>T

NM_000277.2(PAH):c.386A>T (p.Asp129Val)

NC_000012.12:g.102877517T>A

CM000674.2:g.102877517T>A

NC_000012.11:g.103271295T>A

CM000674.1:g.103271295T>A

NC_000012.10:g.101795425T>A

NG_008690.1:g.45086A>T

NG_008690.2:g.85894A>T

ENST00000553106.6:c.386A>T

ENST00000307000.7:c.371A>T

ENST00000549111.5:n.482A>T

ENST00000550978.6:c.370A>T

ENST00000551337.5:c.386A>T

ENST00000551988.5:n.475A>T

ENST00000553106.5:c.386A>T

NM_000277.1:c.386A>T

NM_001354304.1:c.386A>T

NM_000277.3:c.386A>T

NM_001354304.2:c.386A>T

Uncertain Significance

Met criteria codes **3**

PP3

PM2

PM5

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.386A>T (p.Asp129Val) variant in PAH has not been reported in the literature to our knowledge. This variant is absent from 1000G, ESP, ExAC and gnomAD (PM2). A deleterious effect is predicted in SIFT, Polyphen-2, MutationTaster, and REVEL=0.986 (PM3). The

(p.Asp129Gly) variant is likely pathogenic by 1 submitter and PAH VCEP (PM5). In summary, this variant meets criteria to be classified as uncertain significance for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM5, PP3.

Met criteria codes

PP3			Deleterious effect predicted in SIFT, Polyphen2, MutationTaster. REVEL=0.986.
PM2			Absent from ExAC, gnomAD, 1000G, ESP.
PM5			(p.Asp129Gly) is likely pathogenic by 1 submitter and PAH VCEP

Curation History



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