

Variant: NM_000277.2(PAH):c.1089delG (p.Lys363Asnfs)

CA229336 [↗](#)

102518 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: f73ce257-c08b-40ce-845f-012c6403252c

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HGVS expressions

NM_000277.2:c.1089delG

NM_000277.2(PAH):c.1089delG (p.Lys363Asnfs)

NC_000012.12:g.102843756del

CM000674.2:g.102843756del

NC_000012.11:g.103237534del

CM000674.1:g.103237534del

NC_000012.10:g.101761664del

NG_008690.1:g.78847del

NG_008690.2:g.119655del

NM_000277.1:c.1089del

NM_000277.2:c.1089del

NM_001354304.1:c.1089del

NM_000277.3:c.1089del

ENST00000307000.7:c.1074del

ENST00000549247.6:n.848del

ENST00000551114.2:n.751del

ENST00000553106.5:c.1089del

ENST00000635477.1:n.193del

ENST00000635528.1:n.604del

Pathogenic

Met criteria codes **3**

PVS1

PP4

PM2

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: PVS1: Frameshift variant; PM2: Absent from ExAC, gnomAD, 1000G, ESP; PP4: Detected in a patient with Classical PKU. (PMID:8659548). 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: (PVS1, PM2, PP4).

Met criteria codes

PVS1			Frameshift variant
PP4			Detected in a patient with Classical PKU. K363fsdelG detected on 1 chromosome in a patient with Classical PKU. Blood phenylalanine concentrations >1,200 umol/liter, normal blood tyrosine concentrations, and large concentrations of phenylalanine metabolites in urine. PubMed:8659548 
PM2			Absent from ExAC, gnomAD, 1000G, ESP

Curation History



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