

Variant: *NM\_000277.2(PAH):c.1089delG (p.Lys363Asnfs)*

Version: 1.0

CA229336 [↗](#)

102518 (ClinVar) [↗](#)

**Gene:** PAH (HGNC:5053)

**Condition:** phenylketonuria (MONDO:0009861)

**Inheritance Mode:** Autosomal recessive inheritance

**UID:** f73ce257-c08b-40ce-845f-012c6403252c

**Approved on:** 2018-08-10

**Published on:** 2019-08-17

### 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

ENST00000553106.6:c.1089del

ENST00000307000.7:c.1074del

ENST00000549247.6:n.848del

ENST00000551114.2:n.751del

ENST00000553106.5:c.1089del

ENST00000635477.1:c.193del

ENST00000635528.1:n.604del

NM\_000277.1:c.1089del

NM\_000277.2:c.1089del

NM\_001354304.1:c.1089del

NM\_000277.3:c.1089del

NM\_001354304.2:c.1089del

**Pathogenic**

Met criteria codes **3**

PP4 PM2 PVS1

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

<b>PP4</b>	✓	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. <a href="#">PubMed:8659548</a>
<b>PM2</b>	✓	Absent from ExAC, gnomAD, 1000G, ESP
<b>PVS1</b>	✓	Frameshift variant

#### Curation History [↗](#)

Showing 1 to 1 of 1 rows

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