The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

# *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
**UUID:** 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**

| | | |
|---|---|---|
| **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 |
| **PVS1** | ✔ | Frameshift variant |

Curation History

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