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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
Approved on: 2018-08-10
Published on: 2018-12-10

HGVS expressions

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

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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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000277.2(PAH):c.1089delG (p.Lys3...	Pathogenic	Phenylketonuria	2019-08-17	1.0	-	PAH

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