

Variant: *NM_000277.3:c.344_347del*

Version: 1.0

CA229518 [↗](#)

102652 (ClinVar) [↗](#)

Gene: [PAH](#)

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 558984a6-51b6-4793-b0f2-86e7c5b8d83c

Approved on: 2022/10/14

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HGVS expressions

NM_000277.3:c.344_347del

NC_000012.12:g.102894744_102894747del

CM000674.2:g.102894744_102894747del

NC_000012.11:g.103288522_103288525del

CM000674.1:g.103288522_103288525del

NC_000012.10:g.101812652_101812655del

NG_008690.1:g.27860_27863del

NG_008690.2:g.68668_68671del

ENST00000553106.6:c.344_347del

ENST00000307000.7:c.329_332del

ENST00000546844.1:c.344_347del

ENST00000548928.1:n.266_269del

ENST00000549111.5:n.440_443del

ENST00000550978.6:c.328_331del

ENST00000551337.5:c.344_347del

ENST00000551988.5:n.433_436del

ENST00000553106.5:c.344_347del

NM_000277.1:c.344_347del

NM_000277.2:c.344_347del

NM_001354304.1:c.344_347del

NM_001354304.2:c.344_347del

Pathogenic

Met criteria codes **4**

PM3_Supporting

PP4

PM2

PVS1

Evidence Links **0**

Expert Panel

[Phenylketonuria VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Phenylketonuria VCEP

This c.344_347del (p.Lys115fs) variant in PAH was detected with the c.1315+1G>A pathogenic variant and the c.1066-14C>G likely pathogenic variant in multiple patients with PKU, phasing was not available (PMID: 11207989, 28676969, 25551302). This variant was absent in population databases. This is a frameshift variant in exon 3 of 13 coding exons with termination at position 194 predicted to undergo nonsense mediated decay. In summary, this variant meets criteria to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PM2, PM3_Supporting, PP4.

Met criteria codes

PM3_Supporting			This variant was detected with the c.1315+1G>A pathogenic variant (no phasing available) PMID: 25551302. It was detected with the likely pathogenic variant c.1066-14C>G. PMID: 11207989. points=0.75.
PP4			Variant was detected in multiple patients with PKU. PMID: 11207989, 28676969, 25551302.
PM2			This variant is absent from population databases gnomAD and ExAC
PVS1			Frameshift variant predicted to undergo NMD. Termination occurs at position 194 (115+79). Located in exon 3 out of 13 coding exons, not in the last 50bp of exon 12.

Curation History



 

 

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