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_000277.1(PAH):c.111_112insG (p.Ile38Aspfs)

Version: 1.0

CA229354

102531 (ClinVar)

Gene: PAH
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: b99c1dbd-f61d-48b6-b621-b15e0731824b
Approved on: 2019/4/3
Published on: 2019/8/16

HGVS expressions

NM_000277.1(PAH):c.111_112insG (p.Ile38Aspfs)
NM_000277.1:c.111_112insG
NC_000012.12:g.102912848dup
CM000674.2:g.102912848dup
NC_000012.11:g.103306626dup
CM000674.1:g.103306626dup
NC_000012.10:g.101830756dup
NG_008690.1:g.9755dup
NG_008690.2:g.50563dup
ENST00000553106.6:c.111dup
ENST00000307000.7:c.96dup
ENST00000546844.1:c.111dup
ENST00000548677.2:n.198dup
ENST00000548928.1:n.33dup
ENST00000549111.5:n.207dup
ENST00000550978.6:c.95dup
ENST00000551337.5:c.111dup
ENST00000551988.5:n.200dup
ENST00000553106.5:c.111dup
ENST00000635500.1:n.79dup
NM_000277.1:c.111dup
NM_000277.2:c.111dup
NM_001354304.1:c.111dup
NM_000277.3:c.111dup
NM_001354304.2:c.111dup

Pathogenic

Met criteria codes 3

PVS1 PM2 PP4

Evidence Links 1

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.112dupG variant in PAH has been previously reported as a single variant (no second mutation detected) in 1 Chinese patient with classic PKU (PMID: 16256386); BH4 deficiency was not excluded (PP4). The variant is a frameshift variant occurring in exon 2 of 13 in the in the canonical transcript of PAH, a gene fulfilling the most recent criteria for LOF being a known disease mechanism (see PMID: 30192042) (PVS1). It is absent from control databases including ethnically matched individuals, including gnomAD/ExAC, 1000 Genomes, and ESP (PM2). In summary, this variant meets criteria to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PP4, PM2.

Met criteria codes

PVS1	✔	The variant is a frameshift variant occurring in exon 2 of 13 in the in the canonical transcript of PAH, a gene fulfilling the most recent criteria for LOF being a known disease mechanism (see PMID: 30192042) (PVS1).
PM2	✔	It is absent from control databases including ethnically matched individuals, including gnomAD/ExAC, 1000 Genomes, and ESP (PM2).
PP4	✔	The c.112dupG variant in PAH has been previously reported as a single variant (no second mutation detected) in 1 Chinese case with classic PKU (PMID: 16256386); BH4 deficiency was not excluded (PP4). It has also been noted in ClinVar (Variant ID 108267), without further information, and no assertion is provided re its classification.

The c.112dupG variant in PAH has been previously reported as a single variant (no second mutation detected) in 1 Chinese case with classic PKU (PMID: 16256386); BH4 deficiency was not excluded (PP4). PubMed:16256386

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.

¤ Powered by BCM's Genboree.