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.299A>G (p.His100Arg)

CA6748985

306914 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: a445bf0d-e08f-4300-b282-53c974544d50
Approved on: 2018-08-10
Published on: 2018-12-10

HGVS expressions

NM_000277.2(PAH):c.299A>G (p.His100Arg)
NM_000277.2:c.299A>G
NC_000012.12:g.102894788T>C
CM000674.2:g.102894788T>C
NC_000012.11:g.103288566T>C
CM000674.1:g.103288566T>C
NC_000012.10:g.101812696T>C
NG_008690.1:g.27815A>G
NG_008690.2:g.68623A>G
NM_000277.1:c.299A>G
NM_001354304.1:c.299A>G
NM_000277.3:c.299A>G
ENST00000307000.7:c.284A>G
ENST00000546844.1:c.299A>G
ENST00000548928.1:n.221A>G
ENST00000549111.5:n.395A>G
ENST00000550978.6:n.283A>G
ENST00000551337.5:c.299A>G
ENST00000551988.5:n.388A>G
ENST00000553106.5:c.299A>G

Uncertain Significance

Met criteria codes 3

BP4 PP4 PM3

Not Met criteria codes 1

PM2

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: BP4: In silico overwhelmingly predict benign. REVEL = 0.553; PP4: Detected in a patient with non PKU hyperphe (PMID:11244681); PM3: H100R detected with IVS10 (PMID:11244681). In summary this variant meets criteria to be classified as uncertain significance for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (BP4, PP4, PM3).

Met criteria codes

BP4	✔	In silico overwhelmingly predict benign. REVEL = 0.553
PP4	✔	Detected in a patient with non PKU hyperphe We evaluated the clinical data obtained from a multidisciplinary trial of 11 patients with PKU/HPA. H1OOR detected. PubMed:11244681
PM3	✔	H100R detected with IVS10 H100R detected with IVS10. PubMed:11244681

Not Met criteria codes

PM2	✖	> than the PAH specific PM2 specification of AF=0.0002 (0.02%).

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000277.2(PAH):c.299A>G (p.His10...	Uncertain Significance	Phenylketonuria	2019-04-06	1.0	-	PAH

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.