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
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HGVS expressions

NM_000277.2:c.299A>G
NM_000277.2(PAH):c.299A>G (p.His100Arg)
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

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene

No matching records found