

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

Version: 1.0

CA6748985 [↗](#)

306914 (ClinVar) [↗](#)

Gene: PAH (HGNC:5053)

Condition: phenylketonuria (MONDO:0009861)

Inheritance Mode: Autosomal recessive inheritance

UID: a445bf0d-e08f-4300-b282-53c974544d50

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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 PM3 PP4

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
PM3	✓	H100R detected with IVS10 H100R detected with IVS10. PubMed:11244681
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. H100R detected. PubMed:11244681

Not Met criteria codes

PM2	✗	> than the PAH specific PM2 specification of AF=0.0002 (0.02%).
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Curation History [↗](#)

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