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Variant: NM_000277.2(PAH):c.907T>C (p.Ser303Pro)

Version: 1.0

CA229841

102889 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: f8838359-9b6b-4ebd-bdf6-fbd68f1086cb
Approved on: 2019-04-03
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HGVS expressions

NM_000277.2(PAH):c.907T>C (p.Ser303Pro)
NM_000277.2:c.907T>C
NC_000012.12:g.102851692A>G
CM000674.2:g.102851692A>G
NC_000012.11:g.103245470A>G
CM000674.1:g.103245470A>G
NC_000012.10:g.101769600A>G
NG_008690.1:g.70911T>C
NG_008690.2:g.111719T>C
NM_000277.1:c.907T>C
NM_001354304.1:c.907T>C
NM_000277.3:c.907T>C
ENST00000307000.7:c.892T>C
ENST00000549247.6:n.666T>C
ENST00000551114.2:n.569T>C
ENST00000553106.5:c.907T>C
ENST00000635477.1:n.68T>C

Likely Pathogenic

Met criteria codes 4

PP4_Moderate PM3 PM2 PP3

Evidence Links 2

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.907T>C (p.Ser303Pro) variant in PAH has been reported in 1 affected individual (BH4 deficiency excluded). (PP4_Moderate; PMID: 9634518). This variant is absent in population databases (PM2). This variant was detected with p.D282G (LP via PAH workgroup) (PM3; PMID: 16256386). Computational evidence supports a deleterious effect (PP3). In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM3, PP3.

Met criteria codes

PP4_Moderate	✔	Variant identified in a single patient. BH4 defects excluded with urine pterins and DHPR activity in all patients. PMID: 9634518
		Variant identified in a single patient. BH4 defects excluded with urine pterins and DHPR activity in all patients. PubMed:9634518
PM3	✔	Identified in trans with D282G (LP via PAH workgroup) PMID: 16256386
		Identified in a single patient with classic PKU, BH4 defect not excluded, in trans with D282G. PubMed:16256386
PM2	✔	Variant is absent from population databases.
PP3	✔	REVEL Score 0.993, all in silico algorithms agree on deleterious effect

Curation History

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