Variant: NM_000277.3:c.901C>T

Version: 1.0

CA16020888

805824 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 50df6b07-c506-4121-b3c1-dd5a327ad11a
Approved on: 2019-04-04
Published on: 2019-08-16

HGVS expressions

NM_000277.3:c.901C>T
NC_000012.12:g.102851698G>A
CM000674.2:g.102851698G>A
NC_000012.11:g.103245476G>A
CM000674.1:g.103245476G>A
NC_000012.10:g.101769606G>A
NG_008690.1:g.70905C>T
NG_008690.2:g.111713C>T
ENST00000553106.6:c.901C>T
ENST00000307000.7:c.886C>T
ENST00000549247.6:n.660C>T
ENST00000551114.2:n.563C>T
ENST00000553106.5:c.901C>T
ENST00000635477.1:c.62C>T
NM_000277.1:c.901C>T
NM_000277.2:c.901C>T
NM_001354304.1:c.901C>T
NM_001354304.2:c.901C>T

Pathogenic

Met criteria codes 3

PP4 **PM2** **PVS1**

Not Met criteria codes 1

PM3

Evidence Links 1

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.901C>T (p.Q301*) variant in PAH has been reported in at least one patient with PKU (PMID: 20140859). This is a nonsense variant that occurs in exon 9 of PAH, predicted to undergo nonsense mediated decay with the truncated region critical to protein function. This

variant is absent from population databases, including: 1000 Genomes, ESP, and gnomAD. In summary, this variant meets criteria to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PM2, PP4.

Met criteria codes

PP4	✓	Reported in a cohort of Chinese PKU patients. Article in Chinese. Article in Chinese. All of the 13 exons and flanking introns of the PAH gene from 99 patients with PKU were amplified by polymerase chain reaction and analyzed by single strand conformation polymorphism (SSCP), denaturing high performance liquid chromatography (DHPLC) and DNA sequencing. Six novel mutations (IVS3nt+1g--> a, A165D, Q301X, G344D, P362L and R413G) were identified. PubMed:20140859
PM2	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

PM3	✖	Patient genotype not available.

Curation History

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