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Variant: NM_000277.1(PAH):c.1046C>A (p.Ser349Ter)

Version: 1.0

CA229300

102491 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: bc844832-84f0-4266-a9c8-5102faa58165
Approved on: 2020/4/13
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HGVS expressions

NM_000277.1:c.1046C>A
NM_000277.1(PAH):c.1046C>A (p.Ser349Ter)
NC_000012.12:g.102844355G>T
CM000674.2:g.102844355G>T
NC_000012.11:g.103238133G>T
CM000674.1:g.103238133G>T
NC_000012.10:g.101762263G>T
NG_008690.1:g.78248C>A
NG_008690.2:g.119056C>A
ENST00000553106.6:c.1046C>A
ENST00000307000.7:c.1031C>A
ENST00000549247.6:n.805C>A
ENST00000551114.2:n.708C>A
ENST00000553106.5:c.1046C>A
ENST00000635477.1:c.150C>A
ENST00000635528.1:n.561C>A
NM_000277.2:c.1046C>A
NM_001354304.1:c.1046C>A
NM_000277.3:c.1046C>A
NM_001354304.2:c.1046C>A

Likely Pathogenic

Met criteria codes 2

PVS1 PM2

Evidence Links 0

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.1046C>A (p.Ser349Ter) variant in PAH is a null variant (nonsense variant) in exon 10 of 13 in a gene where LOF is a known mechanism of disease, leading to premature truncation and NMD (PVS1). It is absent from ethnically diverse control databases, including

gnomAD/ExAC, 1000 Genomes, and ESP (PM2). It has previously been reported in one classic PKU case per the BioPKU database (PMID: 24939588), without additional information. Classification: Likely Pathogenic Supporting criteria: PVS1, PM2

Met criteria codes

PVS1	✔	The c.976del (p.Trp326GlyfsTer15) variant in PAH is a null variant (frameshift variant) in a gene where LOF is a known mechanism of disease, leading to premature truncation and NMD (PVS1).
PM2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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