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Variant: NM_000277.1(PAH):c.781C>T (p.Arg261Ter)

CA229757

610 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: a79e0701-cceb-4823-a816-a3acbb89f1b7
Approved on: 2018-08-05
Published on: 2018-12-10

HGVS expressions

NM_000277.1:c.781C>T
NM_000277.1(PAH):c.781C>T (p.Arg261Ter)
NC_000012.12:g.102852876G>A
CM000674.2:g.102852876G>A
NC_000012.11:g.103246654G>A
CM000674.1:g.103246654G>A
NC_000012.10:g.101770784G>A
NG_008690.1:g.69727C>T
NG_008690.2:g.110535C>T
NM_000277.2:c.781C>T
NM_001354304.1:c.781C>T
NM_000277.3:c.781C>T
ENST00000307000.7:c.766C>T
ENST00000549247.6:n.540C>T
ENST00000553106.5:c.781C>T

Pathogenic

Met criteria codes 3

PM3 PVS1 PP4

Evidence Links 2

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: PVS1: Nonsense variant; PP4: Detected in 2 unrelated patients (PMID:1682234); PM3: Detected with H170D, pathogenic (PMID:11385716). In summary this variant meets criteria to be classified as pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PVS1, PP4, PM3).

Met criteria codes

PM3	✔	Detected with H170D, pathogenic

Patient genotype: H170D (VarID 92744, Pathogenic) /R261X PubMed:11385716

PVS1 ✔ Nonsense variant

PP4 ✔ Detected in 2 unrelated patients

A new mutation (CGA to TGA) in codon 261 of exon 7 of the phenylalanine hydroxylase gene transforms Arg261 to a stop codon in two unrelated patients of German and Turkish origin. PubMed:1682234

Curation History

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