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Variant: NM_000277.2(PAH):c.727C>T (p.Arg243Ter)

CA220585

588 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 15418e53-7ae7-4fc4-8f8d-246d5db75696
Approved on: 2018-08-07
Published on: 2018-12-10

HGVS expressions

NM_000277.2(PAH):c.727C>T (p.Arg243Ter)
NM_000277.2:c.727C>T
NC_000012.12:g.102852930G>A
CM000674.2:g.102852930G>A
NC_000012.11:g.103246708G>A
CM000674.1:g.103246708G>A
NC_000012.10:g.101770838G>A
NG_008690.1:g.69673C>T
NG_008690.2:g.110481C>T
NM_000277.1:c.727C>T
NM_001354304.1:c.727C>T
NM_000277.3:c.727C>T
ENST00000307000.7:c.712C>T
ENST00000549247.6:n.486C>T
ENST00000553106.5:c.727C>T

Pathogenic

Met criteria codes 2

PS3 PVS1

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: PVS1: nonsense variant; PS3: <1% of normal PAH activity (PMID:2014036). 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, PS3).

Met criteria codes

PS3	✔	<1% of normal PAH activity

R243X mutant PAH activity <1 % of normal level. PubMed:2014036

PVS1		✔	nonsense variant

Curation History

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