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Variant: NM_000277.2(PAH):c.47_48delCT (p.Ser16Terfs)

CA229574

102696 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 88d14684-a7c2-4bfb-9ec0-2e5bc851b285
Approved on: 2018-08-10
Published on: 2018-12-10

HGVS expressions

NM_000277.2:c.47_48delCT
NM_000277.2(PAH):c.47_48delCT (p.Ser16Terfs)
NC_000012.12:g.102917087_102917088del
CM000674.2:g.102917087_102917088del
NC_000012.11:g.103310865_103310866del
CM000674.1:g.103310865_103310866del
NC_000012.10:g.101834995_101834996del
NG_008690.1:g.5519_5520del
NG_008690.2:g.46327_46328del
NM_000277.1:c.47_48del
NM_000277.2:c.47_48del
NM_001354304.1:c.47_48del
NM_000277.3:c.47_48del
ENST00000307000.7:c.-101_-100del
ENST00000546844.1:c.47_48del
ENST00000547319.1:n.358_359del
ENST00000549111.5:n.143_144del
ENST00000550978.6:n.31_32del
ENST00000551337.5:c.47_48del
ENST00000551988.5:n.136_137del
ENST00000553106.5:c.47_48del
ENST00000635500.1:n.29-4186_29-4185del

Pathogenic

Met criteria codes 4
PM3 PM2 PVS1 PP4

Not Met criteria codes 1
PP3

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: PM2: ExAC:8.238e-06; gnomAD:0.000004062; 1000G + ESP: absent; PVS1: Null variant- frameshift. Subject to nonsense mediated decay.; PM3: found in trans with L48S (VarID608, Pathogenic) (PMID:8535445); PP4: 47delCT found in 1 patient with moderate PKU. BH4 deficiency not ruled out. (PMID:8535445). 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: (PM2, PVS1, PM3, PP4).

Met criteria codes

PM3	✔	found in trans with L48S (VarID608, Pathogenic) 47delCT found in trans with L48S in Family 10. PubMed:8535445
PM2	✔	ExAC:8.238e-06; gnomAD:0.000004062; 1000G + ESP: absent
PVS1	✔	Null variant- frameshift. Subject to nonsense mediated decay.
PP4	✔	47delCT found in 1 patient with moderate PKU. BH4 deficiency not ruled out. 42 families with untreated PKU. 47delCT found in Family 10 on 1 allele in trans with L48S. PubMed:8535445

Not Met criteria codes

PP3	✖	Disease causing in MutationTaster

Curation History

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