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Variant: NM_000277.2(PAH):c.127G>T (p.Glu43Ter)

Version: 1.0

CA16020729

557124 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: d224fcf5-f7be-4a92-b876-72d31e405d4f
Approved on: 2019-04-03
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HGVS expressions

NM_000277.2(PAH):c.127G>T (p.Glu43Ter)
NM_000277.2:c.127G>T
NC_000012.12:g.102912832C>A
CM000674.2:g.102912832C>A
NC_000012.11:g.103306610C>A
CM000674.1:g.103306610C>A
NC_000012.10:g.101830740C>A
NG_008690.1:g.9771G>T
NG_008690.2:g.50579G>T
NM_000277.1:c.127G>T
NM_001354304.1:c.127G>T
NM_000277.3:c.127G>T
ENST00000307000.7:c.112G>T
ENST00000546844.1:c.127G>T
ENST00000548677.2:n.214G>T
ENST00000548928.1:n.49G>T
ENST00000549111.5:n.223G>T
ENST00000550978.6:n.111G>T
ENST00000551337.5:c.127G>T
ENST00000551988.5:n.216G>T
ENST00000553106.5:c.127G>T
ENST00000635500.1:n.95G>T

Pathogenic

Met criteria codes 3

PP4 PM2 PVS1

Evidence Links 1

Expert Panel

Phenylketonuria VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.127G>T (p.Glu43*) variant is a nonsense variant in exon 2 of 13 in PAH, predicted to undergo nonsense mediated decay with the truncated region critical to protein function. It has been reported in 2 individuals with PKU (BH4 deficiency not excluded). (PP4; PMID: 26503515). This variant is absent from population databases (PM2). 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	✔	Variant is reported in 2 patients from mainland China in a cohort study. BH4 deficiency not ruled out. PubMed:26503515
PM2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1	✔	Nonsense variant in exon 2 of 13, predicted to undergo nonsense mediated decay with the truncated region critical to protein function

Curation History

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