The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000277.2(PAH):c.1024delG (p.Ala342Hisfs)

Version: 1.0

CA229279

102475 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: 4cc2d322-d860-4209-b3df-63e58bac3b07
Approved on: 2018-08-10
Published on: 2019-08-17

HGVS expressions

NM_000277.2:c.1024delG
NM_000277.2(PAH):c.1024delG (p.Ala342Hisfs)
NC_000012.12:g.102844378del
CM000674.2:g.102844378del
NC_000012.11:g.103238156del
CM000674.1:g.103238156del
NC_000012.10:g.101762286del
NG_008690.1:g.78226del
NG_008690.2:g.119034del
ENST00000553106.6:c.1024del
ENST00000307000.7:c.1009del
ENST00000549247.6:n.783del
ENST00000551114.2:n.686del
ENST00000553106.5:c.1024del
ENST00000635477.1:c.128del
ENST00000635528.1:n.539del
NM_000277.1:c.1024del
NM_000277.2:c.1024del
NM_001354304.1:c.1024del
NM_000277.3:c.1024del
NM_001354304.2:c.1024del

Pathogenic

Met criteria codes 3

PVS1 PP4_Moderate PM2

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: Absent from ExAC, gnomAD, 1000G, ESP; PVS1: Frameshift variant; PP4_Moderate: Reported in patients with PAH deficiency. Bh4 defects excluded. (PMID:9634518). 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, PP4_Moderate).

Met criteria codes

PVS1	✔	Frameshift variant
PP4_Moderate	✔	Reported in patients with PAH deficiency. Bh4 defects excluded. A342fsdelG detected. PAH deficiency had been assessed after exclusion of a defect in tetrahydrobiopterin metabolism. **PubMed:9634518**
PM2	✔	Absent from ExAC, gnomAD, 1000G, ESP

Curation History

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