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.806delT (p.Ile269Thrfs)

Version: 1.0

CA229778

102844 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: f5d8dc3f-dba0-4dc9-98cd-4a22b6a11a83
Approved on: 2018-08-10
Published on: 2019-08-17

HGVS expressions

NM_000277.2:c.806delT
NM_000277.2(PAH):c.806delT (p.Ile269Thrfs)
NC_000012.12:g.102852851del
CM000674.2:g.102852851del
NC_000012.11:g.103246629del
CM000674.1:g.103246629del
NC_000012.10:g.101770759del
NG_008690.1:g.69752del
NG_008690.2:g.110560del
ENST00000553106.6:c.806del
ENST00000307000.7:c.791del
ENST00000549247.6:n.565del
ENST00000553106.5:c.806del
NM_000277.1:c.806del
NM_000277.2:c.806del
NM_001354304.1:c.806del
NM_000277.3:c.806del
NM_001354304.2:c.806del

Pathogenic

Met criteria codes 3

PM2 PP4 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: Frameshift variant; PM2: Extremely low frequency. gnomAD MAF=0.00007.; PP4: Detected in a PKU patient. BH4 deficiency not assessed. (PMID:9012412). 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, PM2, PP4).

Met criteria codes

PM2	✔	Extremely low frequency. gnomAD MAF=0.00007.
PP4	✔	Detected in a PKU patient. BH4 deficiency not assessed.
		I269fsdelT detected on 1 PKU chromosome. PubMed:9012412
PVS1	✔	Frameshift variant

Curation History

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