

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

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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**

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

Curation History 	
 Showing 1 to 1 of 1 rows	

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