

Variant: NM_000277.2(PAH):c.503delA (p.Tyr168Serfs)

CA229588 [↗](#)

102705 (ClinVar) [↗](#)

Gene: PAH ([HGNC:5053](#))

Condition: phenylketonuria ([MONDO:0009861](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 7a494430-377e-4473-a5f7-e0698ec51cab

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HGVS expressions

NM_000277.2:c.503delA

NM_000277.2(PAH):c.503delA (p.Tyr168Serfs)

NC_000012.12:g.102866602del

CM000674.2:g.102866602del

NC_000012.11:g.103260380del

CM000674.1:g.103260380del

NC_000012.10:g.101784510del

NG_008690.1:g.56001del

NG_008690.2:g.96809del

NM_000277.1:c.503del

NM_000277.2:c.503del

NM_001354304.1:c.503del

NM_000277.3:c.503del

ENST00000307000.7:c.488del

ENST00000549111.5:n.599del

ENST00000551988.5:n.530+10860del

ENST00000553106.5:c.503del

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

PAH-specific ACMG/AMP criteria applied: PVS1: Frameshift variant; PM2: Extremely low frequency. ExAC MAF: 0.00001.; PP4: Detected in PKU patient in international phase II clinical trial for sapropterin. (PMID:23430918). 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

PP4



Detected in PKU patient in international phase II clinical trial for sapropterin.

c.503delA (p.Y168>Sfs) detected in study. [PubMed:23430918](#)

PM2



Extremely low frequency. ExAC MAF: 0.00001.

PVS1



Frameshift variant

Curation History

▼

▼

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