

Variant: *NM\_000277.2(PAH):c.503delA (p.Tyr168Serfs)*

Version: 1.0

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

ENST00000553106.6:c.503del

ENST00000307000.7:c.488del

ENST00000549111.5:n.599del

ENST00000551988.5:n.530+10860del

ENST00000553106.5:c.503del

NM\_000277.1:c.503del

NM\_000277.2:c.503del

NM\_001354304.1:c.503del

NM\_000277.3:c.503del

NM\_001354304.2:c.503del

**Pathogenic**

Met criteria codes **3**

PP4 PVS1 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. 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

<b>PP4</b>	✓	Detected in PKU patient in international phase II clinical trial for sapropterin. c.503delA (p.Y168>Sfs) detected in study. <a href="#">PubMed:23430918</a>
<b>PVS1</b>	✓	Frameshift variant
<b>PM2</b>	✓	Extremely low frequency. ExAC MAF: 0.00001.

#### Curation History [↗](#)

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