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.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
Approved on: 2018-08-10
Published on: 2019-08-17

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 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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