

Variant: *NM_000277.3(PAH):c.163_165del (p.Phe55del)*

Version: 1.0

CA229450 [↗](#)

102603 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: b8f27de0-e08a-4c93-ad6a-2192544aaacc

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HGVS expressions

NM_000277.3:c.163_165del

NM_000277.3(PAH):c.163_165del (p.Phe55del)

NC_000012.12:g.102912794_102912796del

CM000674.2:g.102912794_102912796del

NC_000012.11:g.103306572_103306574del

CM000674.1:g.103306572_103306574del

NC_000012.10:g.101830702_101830704del

NG_008690.1:g.9807_9809del

NG_008690.2:g.50615_50617del

ENST00000553106.6:c.163_165del

ENST00000307000.7:c.148_150del

ENST00000546844.1:c.163_165del

ENST00000548677.2:n.250_252del

ENST00000548928.1:n.85_87del

ENST00000549111.5:n.259_261del

ENST00000550978.6:c.147_149del

ENST00000551337.5:c.163_165del

ENST00000551988.5:n.252_254del

ENST00000553106.5:c.163_165del

ENST00000635500.1:n.131_133del

NM_000277.1:c.163_165del

NM_000277.2:c.163_165del

NM_001354304.1:c.163_165del

NM_001354304.2:c.163_165del

Likely Pathogenic

Met criteria codes **4**

PP4 **PM2** **PM3** **PM4**

Not Met criteria codes **1**

BP3

Evidence Links **0**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Phenylketonuria VCEP

This c.163_165del (p.Phe55del) variant in PAH was detected in trans with the c.168+5G>C and p.Phe39Leu pathogenic variants in multiple individuals with PKU (PMID:28676969, 17513426). This variant is absent in population databases. This in-frame variant is not found in a repetitive region and does not cause a stop-loss. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PM2, PM3, PP4, PM4

Met criteria codes

PP4			This variant was detected in multiple individuals affected with PKU (PMID:28676969, 17513426, 27469133)
PM2			This variant is absent from population databases gnomAD and ExAC.
PM3			This variant was detected in trans with the pathogenic variant c.168+5G>C in a patient with PKU (PMID:27469133) and with the pathogenic variant p.Phe39Leu patient mPKU (PMID: 17513426). points=1.
PM4			This in-frame variant is not found in a repetitive region without a known function. It i does not cause a stop loss variant.

Not Met criteria codes

BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History

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