

Variant: *NM_000277.1:c.48dup*

Version: 1.0

CA16020720 [↗](#)

805804 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 1eae3b8a-7f13-4ed6-a03c-e9edf537ff1d

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HGVS expressions

NM_000277.1:c.48dup

NC_000012.12:g.102917083dup

CM000674.2:g.102917083dup

NC_000012.11:g.103310861dup

CM000674.1:g.103310861dup

NC_000012.10:g.101834991dup

NG_008690.1:g.5520dup

NG_008690.2:g.46328dup

ENST00000553106.6:c.48dup

ENST00000307000.7:c.-100dup

ENST00000546844.1:c.48dup

ENST00000547319.1:n.359dup

ENST00000549111.5:n.144dup

ENST00000550978.6:c.32dup

ENST00000551337.5:c.48dup

ENST00000551988.5:n.137dup

ENST00000553106.5:c.48dup

ENST00000635500.1:n.29-4185dup

NM_000277.2:c.48dup

NM_001354304.1:c.48dup

NM_000277.3:c.48dup

NM_001354304.2:c.48dup

Pathogenic

Met criteria codes **4**

PM3_Supporting

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

Pathogenic: The c.48dup (p.D17*) variant has been reported in one individual with classic PKU, who carried a second pathogenic variant in PAH (p.L48S) (PM3, PP4; PMID: 18299955). This variant is absent from gnomAD, 1000 Genomes, and ESP databases (PM2). In summary, this variant meets criteria to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PM3-supporting, PM2, PP4.

Met criteria codes

PM3_Supporting			<p>This variant was detected in a patient with classic PKU phenotype, who carried a second PAH variant (p.L48S). PMID: 18299955. Parental analysis not reported.</p> <p>This variant was detected in a patient with classic PKU phenotype, who carried a second PAH variant (p.L48S). PubMed:18299955 </p>
PP4			<p>This variant was detected in a patient with classic PKU phenotype. The patients were tested for the exclusion of lacking the cofactor BH4 for the differential characterization of the cause for the absent of PAH. PMID: 18299955</p> <p>This variant was detected in a patient with classic PKU phenotype. The patients were tested for the exclusion of lacking the cofactor BH4 for the differential characterization of the cause for the absent of PAH. PubMed:18299955 </p>
PM2			<p>absent from gnomAD</p>
PVS1			<p>No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline</p>

Curation History

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Showing 1 to 1 of 1 rows

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