

Variant: *NM_000277.1:c.197_204del*

Version: 1.0

CA16021007 [↗](#)

1693244 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: af0e89ef-9142-4204-831b-dc5c37c6c154

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HGVS expressions

NM_000277.1:c.197_204del

NC_000012.12:g.102894883_102894890del
CM000674.2:g.102894883_102894890del
NC_000012.11:g.103288661_103288668del
CM000674.1:g.103288661_103288668del
NC_000012.10:g.101812791_101812798del
NG_008690.1:g.27713_27720del
NG_008690.2:g.68521_68528del
ENST00000553106.6:c.197_204del
ENST00000307000.7:c.182_189del
ENST00000546844.1:c.197_204del
ENST00000548677.2:n.284_291del
ENST00000548928.1:n.119_126del
ENST00000549111.5:n.293_300del
ENST00000550978.6:c.181_188del
ENST00000551337.5:c.197_204del
ENST00000551988.5:n.286_293del
ENST00000553106.5:c.197_204del
ENST00000635500.1:n.165_172del
NM_000277.2:c.197_204del
NM_001354304.1:c.197_204del
NM_000277.3:c.197_204del
NM_001354304.2:c.197_204del

Pathogenic

Met criteria codes **4**

PP4_Moderate

PVS1

PM3_Supporting

PM2

Evidence Links **1**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.197_204del8 (p.Glu66Alafs*8) variant in PAH has been reported in 1 individual with PAH deficiency (BH4 deficiency excluded, PP4_Moderate; PMID: 21147011). This variant was detected with p.A300S (Pathogenic in ClinVar) (PM3_supporting; PMID: 21147011). It is a frameshift variant predicted to lead to NMD (PVS1). It is absent from gnomAD (PM2). In summary, this variant meets criteria to be classified as pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PVS1, PP4_Moderate, PM3, PM2.

Met criteria codes

PP4_Moderate			c.197-204del8 was detected on 1 patient (called D75X). BH4 defects assessed. PMID: 21147011
			588 hyperphenylalaninemic patients were investigated. Assessment included PAH gene and genes of the BH4 synthesis/recycling pathways (PTS and QDPR). c.197-204del8 was detected on 1 patient (called D75X). PubMed:21147011 
PVS1			Frameshift
PM3_Supporting			Detected with A300S (pathogenic in ClinVar) PMID: 21147011 parental analysis not confirmed
			Allele 1:p.D75X, Allele 2: p.A300S (VarID92751, Pathogenic) PubMed:21147011 
PM2			absent from gnomAD v2.1.1

Curation History

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