

Variant: *NM_000277.1(PAH):c.266dupC (p.Ala90Cysfs)*

Version: 1.0

CA229503 [↗](#)

102642 (ClinVar) [↗](#)

Gene: [PAH](#)

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 576bbc2e-c3f8-4d05-8b80-899d55eae361

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HGVS expressions

NM_000277.1(PAH):c.266dupC (p.Ala90Cysfs)

NM_000277.1:c.266dupC

NM_000277.1:c.264_265insC

NM_000277.1:c.266_267insC

NC_000012.12:g.102894822dup

CM000674.2:g.102894822dup

NC_000012.11:g.103288600dup

CM000674.1:g.103288600dup

NC_000012.10:g.101812730dup

NG_008690.1:g.27782dup

NG_008690.2:g.68590dup

ENST00000553106.6:c.266dup

ENST00000307000.7:c.251dup

ENST00000546844.1:c.266dup

ENST00000548677.2:n.353dup

ENST00000548928.1:n.188dup

ENST00000549111.5:n.362dup

ENST00000550978.6:c.250dup

ENST00000551337.5:c.266dup

ENST00000551988.5:n.355dup

ENST00000553106.5:c.266dup

NM_000277.1:c.266dup

NM_000277.2:c.266dup

NM_001354304.1:c.266dup

NM_000277.3:c.266dup

NM_001354304.2:c.266dup

Pathogenic

Met criteria codes **4**

PM2

PM3

PVS1

PP4_Moderate

Evidence Links **1**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Phenylketonuria VCEP

Variant classified as Pathogenic using the following criteria: PVS1; PM2; PM3; PP4_Moderate. PVS1: frameshift variant in exon 2 of 13, predicted to result in PTC with removal of >10% of the protein and NMD; PM2: absent from ExAC, gnomAD, 1000G, ESP. PP4_Moderate; PM3: c.266_267insC seen on 2 PKU alleles, with BH4 deficiency ruled out (PMID: 21147011). Detected with pathogenic variants IVS12+1G>A and p.I65T (PMID: 9452062); and p.Pro281Leu (Likely Pathogenic per ClinGen PAH VCEP, PMID: 26666653).

Met criteria codes

PM2			absent from ExAC, gnomAD, 1000G, ESP
PM3			found in with IVS12+1G>A (Pathogenic per ClinGen PAH VCEP) and p.I65T (P 10 submitters) parental analysis not reported (PMID: 9452062); with p.Pro281Leu (Likely Pathogenic per ClinGen PAH VCEP) in one French PKU case parental analysis not reported (PMID: 26666653). p.P281R in 2 patients, parental analysis not reported PMID: 21147011
PubMed:21147011 			
PVS1			PVS1: frameshift variant in exon 2 of 13, predicted to result in PTC with removal of >10% of the protein and NMD
PP4_Moderate			c.266_267insC seen on 2 PKU alleles. BH4 deficiency ruled out. Upgraded per ClinGen Metabolic workgroup
588 hyperphenylalaninemic patients were investigated. Assessment included PAH gene and genes of the BH4 synthesis/recycling pathways (PTS and QDPR). c.266_267insC seen in 2 alleles. PubMed:21147011 			

Curation History



 

 

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