

Variant: *NM_000277.3:c.443G>A*

Version: 1.0

CA16020790 [↗](#)

872837 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: ca00cb65-08c6-4727-ade1-dd87efb4f578

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HGVS expressions

NM_000277.3:c.443G>A

NC_000012.12:g.102866662C>T

CM000674.2:g.102866662C>T

NC_000012.11:g.103260440C>T

CM000674.1:g.103260440C>T

NC_000012.10:g.101784570C>T

NG_008690.1:g.55941G>A

NG_008690.2:g.96749G>A

ENST00000553106.6:c.443G>A

ENST00000307000.7:c.428G>A

ENST00000549111.5:n.539G>A

ENST00000551988.5:n.530+10800G>A

ENST00000553106.5:c.443G>A

NM_000277.1:c.443G>A

NM_000277.2:c.443G>A

NM_001354304.1:c.443G>A

NM_001354304.2:c.443G>A

Likely Pathogenic

Met criteria codes **4**

PP4_Moderate

PM3_Supporting

PP3

PM2

Evidence Links **2**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

The c.443G>A (p.Gly148Asp) variant in PAH has been reported in multiple individuals with PKU (BH4 deficiency excluded). (PMID: 21147011, 15589814). This variant is absent in population databases (PM2). This variant was detected with pathogenic variant IVS4nt5G>T (c.441+5G>T). Computational evidence supports a deleterious effect. In summary, this variant meets criteria to be classified as likely pathogenic for PAH. PAH-specific ACMG/AMP criteria applied: PP4_Moderate, PM2, PM3_supporting, PP3.

Met criteria codes

PP4_Moderate			Detected on 2 alleles. Sequencing of the PAH gene and genes of the BH4 synthesis/ recycling pathways were performed. PMID: 21147011
PubMed:15589814  PubMed:21147011 			
PM3_Supporting			Detected with IVS4nt5G>T (c.441+5G>T) pathogenic 5 submitters PMID: 15589814
PP3			Predicted deleterious in SIFT, PolyPhen2, MutationTaster, REVEL=0.944
PM2			Absent from controls in ExAC, gnomAD, 1000 Genomes, or ESP

Curation History



 

 

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