

Variant: NM_000277.2(PAH):c.1055delG (p.Gly352Valfs)

CA229311 [↗](#)

102498 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 65776af0-77ed-41ec-9999-612a44a636b2

Approved on: 2018-08-10

Published on: 2018-12-10

HGVS expressions

NM_000277.2:c.1055delG

NM_000277.2(PAH):c.1055delG (p.Gly352Valfs)

NC_000012.12:g.102844347del

CM000674.2:g.102844347del

NC_000012.11:g.103238125del

CM000674.1:g.103238125del

NC_000012.10:g.101762255del

NG_008690.1:g.78257del

NG_008690.2:g.119065del

NM_000277.1:c.1055del

NM_000277.2:c.1055del

NM_001354304.1:c.1055del

NM_000277.3:c.1055del

ENST00000307000.7:c.1040del

ENST00000549247.6:n.814del

ENST00000551114.2:n.717del

ENST00000553106.5:c.1055del

ENST00000635477.1:n.159del

ENST00000635528.1:n.570del

Pathogenic

Met criteria codes **3**

PVS1

PP4

PM2

Evidence Links **1**

Expert Panel

Phenylketonuria VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Phenylketonuria VCEP

PAH-specific ACMG/AMP criteria applied: PVS1: Frameshift variant; PM2: Extremely low frequency in ExAC, MAF=0.00002.; PP4: Identified in a pair of siblings with PKU. (PMID:7913581). In summary this variant meets criteria to be classified as pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PVS1, PM2, PP4).

Met criteria codes

PVS1			Frameshift variant
PP4			Identified in a pair of siblings with PKU. identified novel mutation (1054/1055delG[352fs]) in a pair of Italian PKU sibs. Each patient had persistent elevation of blood phenylalanine in the absence of treatment and met the differential criteria for PAH deficiency (Scriver et al. 1989). PubMed:7913581 
PM2			Extremely low frequency in ExAC, MAF=0.00002.

Curation History

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