

Variant: *NM\_000277.2(PAH):c.1055delG (p.Gly352Valfs)*

Version: 1.0

CA229311 [↗](#)

102498 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal recessive inheritance

**UID:** 65776af0-77ed-41ec-9999-612a44a636b2

**Approved on:** 2018-08-10

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### *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

ENST00000553106.6:c.1055del

ENST00000307000.7:c.1040del

ENST00000549247.6:n.814del

ENST00000551114.2:n.717del

ENST00000553106.5:c.1055del

ENST00000635477.1:c.159del

ENST00000635528.1:n.570del

NM\_000277.1:c.1055del

NM\_000277.2:c.1055del

NM\_001354304.1:c.1055del

NM\_000277.3:c.1055del

NM\_001354304.2:c.1055del

**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

<b>PVS1</b>	✓	Frameshift variant
<b>PP4</b>	✓	Identified in a pair of siblings with PKU. <hr/> 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). <a href="#">PubMed:7913581</a> 
<b>PM2</b>	✓	Extremely low frequency in ExAC, MAF=0.00002.

#### Curation History

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