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
UUID: 65776af0-77ed-41ec-9999-612a44a636b2
Approved on: 2018-08-10
Published on: 2019-08-17

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

PP4 PM2 PVS1

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

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.
PVS1	✔	Frameshift variant

Curation History

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