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Variant: NM_000277.2(PAH):c.912+1G>A

Version: 1.0

CA220591

92752 (ClinVar)

Gene: PAH (HGNC:5053)
Condition: phenylketonuria (MONDO:0009861)
Inheritance Mode: Autosomal recessive inheritance
UUID: f8b8c165-463e-4138-88c6-316879bdcf67
Approved on: 2018-08-10
Published on: 2019-08-17

HGVS expressions

NM_000277.2:c.912+1G>A
NM_000277.2(PAH):c.912+1G>A
NC_000012.12:g.102851686C>T
CM000674.2:g.102851686C>T
NC_000012.11:g.103245464C>T
CM000674.1:g.103245464C>T
NC_000012.10:g.101769594C>T
NG_008690.1:g.70917G>A
NG_008690.2:g.111725G>A
ENST00000553106.6:c.912+1G>A
ENST00000307000.7:c.897+1G>A
ENST00000549247.6:n.671+1G>A
ENST00000551114.2:n.574+1G>A
ENST00000553106.5:c.912+1G>A
ENST00000635477.1:c.73+1G>A
NM_000277.1:c.912+1G>A
NM_001354304.1:c.912+1G>A
NM_000277.3:c.912+1G>A
NM_001354304.2:c.912+1G>A

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: PM2: Extremely low frequency. ExAC MAF=0.00006.; PVS1: Canonical +1 splice site; PP4: Detected in 5 patients with classical PKU. (PMID:8659548). 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: (PM2, PVS1, PP4).

Met criteria codes

PP4	✔	Detected in 5 patients with classical PKU. IVS8nt1g>a was detected on 5 chromosomes of patients with classical PKU. Blood phenylalanine concentrations >1,200 umol/liter, normal blood tyrosine concentrations, and large concentrations of phenylalanine metabolites in urine. PubMed:8659548
PM2	✔	Extremely low frequency. ExAC MAF=0.00006.
PVS1	✔	Canonical +1 splice site

Curation History

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