

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

CA220591 [↗](#)

92752 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: f8b8c165-463e-4138-88c6-316879bdcf67

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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

NM_000277.1:c.912+1G>A

NM_001354304.1:c.912+1G>A

NM_000277.3: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:n.73+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.
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			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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