

Variant: NM_000277.2(PAH):c.535T>A (p.Tyr179Asn)

CA229613 [↗](#)

102729 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 1530f57a-9096-4f98-a934-15ce8f576c73

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HGVS expressions

NM_000277.2:c.535T>A

NM_000277.2(PAH):c.535T>A (p.Tyr179Asn)

NC_000012.12:g.102855307A>T

CM000674.2:g.102855307A>T

NC_000012.11:g.103249085A>T

CM000674.1:g.103249085A>T

NC_000012.10:g.101773215A>T

NG_008690.1:g.67296T>A

NG_008690.2:g.108104T>A

NM_000277.1:c.535T>A

NM_001354304.1:c.535T>A

NM_000277.3:c.535T>A

ENST00000307000.7:c.520T>A

ENST00000549111.5:n.631T>A

ENST00000551988.5:n.556T>A

ENST00000553106.5:c.535T>A

Likely Pathogenic

Met criteria codes **4**

PP4_Moderate

PP3

PM2

PM3

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: Absent from ExAC, gnomAD, 1000G, ESP; PP3: Deleterious effect predicted in SIFT, Polyphen-2, MutationTaster. REVEL=0.929; PP4_Moderate: Detected in 1 PKU patient, primary BH4 deficiency excluded. Upgraded per ClinGen PAH EP. (PMID:23430918); PM3: Detected with c.1066-11G>A (P) (PMID:23430918). In summary this variant meets criteria to be classified as likely pathogenic for phenylketonuria in an autosomal recessive manner based on the ACMG/AMP criteria applied as specified by the PAH Expert Panel: (PM2, PP3, PP4_Moderate, PM3).

Met criteria codes

PP4_Moderate			<div>Detected in 1 PKU patient, primary BH4 deficiency excluded. Upgraded per ClinGen PAH EP.</div> <div>Detected in 1 PKU patient: previously documented Phe level >360 umol/L and a Phe level >450 umol/L at the screening visit. Patients with a diagnosis of primary BH4 deficiency were excluded from the study.</div> <div>PubMed:23430918 </div>
PP3			Deleterious effect predicted in SIFT, Polyphen-2, MutationTaster. REVEL=0.929
PM2			Absent from ExAC, gnomAD, 1000G, ESP
PM3			<div>Detected with c.1066-11G>A (P)</div> <div>Patient genotype: p.Y179N: c.1066-11G>A PubMed:23430918 </div>

Curation History

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