

Variant: NM_000277.2(PAH):c.526C>T (p.Arg176Ter)

Version: 1.0

CA275338 [↗](#)

102723 (ClinVar) [↗](#)

Gene: PAH (HGNC:5053)

Condition: phenylketonuria (MONDO:0009861)

Inheritance Mode: Autosomal recessive inheritance

UID: 08d51386-af9e-4de1-8e7b-0dc6c2252c91

Approved on: 2018-08-13

Published on: 2019-04-06

HGVS expressions

NM_000277.2:c.526C>T

NM_000277.2(PAH):c.526C>T (p.Arg176Ter)

NC_000012.12:g.102855316G>A

CM000674.2:g.102855316G>A

NC_000012.11:g.103249094G>A

CM000674.1:g.103249094G>A

NC_000012.10:g.101773224G>A

NG_008690.1:g.67287C>T

NG_008690.2:g.108095C>T

NM_000277.1:c.526C>T

NM_001354304.1:c.526C>T

NM_000277.3:c.526C>T

ENST00000307000.7:c.511C>T

ENST00000549111.5:n.622C>T

ENST00000551988.5:n.547C>T

ENST00000553106.5:c.526C>T

Pathogenic

Met criteria codes **4**

PP4_Moderate

PM3_Strong

PM2

PVS1

Evidence Links **2**

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: Nonsense variant; PM2: ExAC MAF: 0.00010; PP4_Moderate: BH4 defect excluded in all patients in Liu 2015. Identified in 6 patients in this study (PMID:10394930; PMID:26600521); PM3_Strong: Identified in 6 patients, in trans with R243Q and R241C (both pathogenic) (PMID:26600521). 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_Moderate, PM3_Strong).

Met criteria codes

PP4_Moderate	✓	BH4 defect excluded in all patients in Liu 2015. Identified in 6 patients in this study Patients with PAH-deficient hyperphenylalaninaemia in 272 independent families (248 PKU and 24 MHP) living in Germany were investigated. R176X was detected on 1 chromosome. PubMed:10394930 BH4 defect excluded in all patients PubMed:26600521
PM3_Strong	✓	Identified in 6 patients, in trans with R243Q and R241C (both pathogenic) Identified in 6 patients, in trans with R243Q and R241C (both pathogenic) PubMed:26600521
PM2	✓	ExAC MAF: 0.00010
PVS1	✓	Nonsense variant

Curation History [↗](#)

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.