

Variant: *NM_001204.7(BMPR2):c.240_241insT (p.Lys81Ter)*

Version: 1.0

CA645293999 [↗](#)

425725 (ClinVar) [↗](#)

Gene: BMPR2 ([HGNC:659](#))

Condition: pulmonary arterial hypertension ([MONDO:0015924](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 3f6b7a5a-1054-4f88-8716-48d26795389c

Approved on: 2024/5/3

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HGVS expressions

NM_001204.7:c.240_241insT

NM_001204.7(BMPR2):c.240_241insT (p.Lys81Ter)

NC_000002.12:g.202464972_202464973insT

CM000664.2:g.202464972_202464973insT

NC_000002.11:g.203329695_203329696insT

CM000664.1:g.203329695_203329696insT

NC_000002.10:g.203037940_203037941insT

NG_009363.1:g.93646_93647insT

ENST00000374580.10:c.240_241insT

ENST00000638587.1:c.165_166insT

ENST00000374574.2:c.240_241insT

ENST00000374580.8:c.240_241insT

ENST00000479069.1:n.147_148insT

NM_001204.6:c.240_241insT

Likely Pathogenic

Met criteria codes **2**

PM2_Supporting

PVS1

Evidence Links **0**

Expert Panel

Pulmonary Hypertension VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Pulmonary Hypertension Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for BMPR2 Version 1.1.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Pulmonary Hypertension VCEP

The NM_001204.7(BMPR2):c.240_241insT (p.Lys81Ter) variant is a DNA single thymine base insertion predicted to cause a frameshift, replacing a lysine residue for a stop codon at position 81. The variant is absent from the gnomAD v.2.1.1 controls and v3.0 (PM2_supporting). c.240_241insT was found in a patient with pulmonary arterial hypertension (PMID:20496075). The variant resides in the second exon of BMPR2 and generates a stop codon that is predicted to cause nonsense-mediated decay (PVS1). In summary, this variant

meets the criteria to be classified as likely pathogenic for pulmonary arterial hypertension based on ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP: PM2_supporting, PVS1 (VCEP specification version v 1.1, 1/18/2024).

Met criteria codes

PM2_Supporting

i

✓

The variant is absent from gnomAD genomes v3.0 and gnomAD exomes v.2.1.1

PVS1

i

✓

The insertion of a single thymine in the exon 2 of BMPR2 replaces the lysine residue in position 81 with a stop codon. The presence of a premature stop codon is predicted to result in a truncated form of the protein that is often non-functional. The mRNA in this case is predicted to undergo nonsense-mediated decay.

Curation History [↗](#)

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