

Variant: *NM_001204.7(BMPR2):c.1151C>T (p.Ala384Val)*

Version: 2.0

CA350341728 [↗](#)

425882 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: f2b50603-89e4-4ca8-a22e-89d70ef18705

Approved on: 2026/4/9

Published on: 2026/4/9

HGVS expressions

NM_001204.7:c.1151C>T

NM_001204.7(BMPR2):c.1151C>T (p.Ala384Val)

NC_000002.12:g.202532607C>T

CM000664.2:g.202532607C>T

NC_000002.11:g.203397330C>T

CM000664.1:g.203397330C>T

NC_000002.10:g.203105575C>T

NG_009363.1:g.161281C>T

ENST00000374580.10:c.1151C>T

ENST00000638587.1:c.1082C>T

ENST00000374574.2:c.1151C>T

ENST00000374580.8:c.1151C>T

NM_001204.6:c.1151C>T

Likely Pathogenic

Met criteria codes 5

PS4_Supporting

PP2

PP3

PM1

PM2_Supporting

Not Met criteria codes 5

BA1

BS1

BP4

PS1

PM5

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 2.0.0

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

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

Evidence submitted by expert panel

Pulmonary Hypertension VCEP

The c.1151C>T (p.Ala384Val) variant is a missense variant located in exon 9 of the BMPR2 gene, predicted to cause substitution of alanine to valine encoding the functionally relevant catalytic kinase domain but without functional evidence indicating critical or non-critical (PM1_moderate). This variant is absent from gnomAD v2.1.1 (controls) and v4.1 (PM2_supporting). BMPR2 has a low rate of benign missense variants and missense variants are a common mechanism of disease (PP2). The REVEL prediction algorithm score is 0.917 and AlphaMissense is 0.9915 indicating pathogenicity (PP3_met). The variant has been reported in one Japanese individual (PMID: 23675998)

and one Chinese individual (PMID: 30578397, PMID: 32634488) with idiopathic PAH (PS4_supporting). PS2 was not assessed due to lack of paternity data. Functional studies have not been conducted for this variant (PS3 not assessed). In summary, this variant meets the criteria to be classified as a likely pathogenic for pulmonary arterial hypertension based on the ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP: PM1_moderate, PM2_supporting, PP2, PP3_supporting (VCEP specification version 2.0, 1/30/2026).

Met criteria codes

PS4_Supporting			The variant has been reported in one Japanese individual (PMID: 23675998) and one Chinese individual (PMID: 30578397, PMID: 32634488) with idiopathic PAH.
PP2			BMPR2 has a z-score of 3.28 for missense variants which is above the threshold of 3.09 (PMID: 40496714) for constraint.
PP3			The REVEL prediction algorithm score is 0.917, AlphaMissense is 0.9915 indicating pathogenicity.
PM1			Variant is located in the conserved catalytic kinase domain but without functional evidence indicating critical or non-critical amino acid residue.
PM2_Supporting			Variant is absent from gnomAD v2.1.1 controls and v4.1.

Not Met criteria codes

BA1			Variant is absent from gnomAD v2.1.1 controls and v4.1.
BS1			Variant is absent from gnomAD v2.1.1 controls and v4.1.
BP4			The REVEL prediction algorithm score is 0.917, AlphaMissense is 0.9915 indicating pathogenicity.
PS1			The same amino acid change has not been reported.
PM5			No other variants at the amino acid residue have been reported.

Curation History 



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