

Variant: *NM_001204.7(BMPR2):c.2352C>T (p.Val784=)*

Version: 1.2

CA2061522 [↗](#)

333647 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 56885255-cd86-4c5d-887f-dfd7f2b7ad29

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HGVS expressions

NM_001204.7:c.2352C>T

NM_001204.7(BMPR2):c.2352C>T (p.Val784=)

NC_000002.12:g.202556017C>T

CM000664.2:g.202556017C>T

NC_000002.11:g.203420740C>T

CM000664.1:g.203420740C>T

NC_000002.10:g.203128985C>T

NG_009363.1:g.184691C>T

ENST00000374580.10:c.2352C>T

ENST00000638587.1:c.2283C>T

ENST00000374574.2:c.1586+3129C>T

ENST00000374580.8:c.2352C>T

NM_001204.6:c.2352C>T

Uncertain Significance

Met criteria codes **3**

BP7

BP4

PM2_Supporting

Not Met criteria codes **4**

BA1

BS1

PS4

PM1

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 NM_001204.7(BMPR2) c.2352C>T variant is a synonymous (silent) variant (p.Val784=). This variant is present in gnomADv2.1.1 controls at a frequency of 0.00002 and gnomADv4.1 at 0.00006 (PM2_supporting). In silico prediction algorithms indicate that the nucleotide is not highly conserved (REVEL score not available; CADD = 1.4; phyloP = 0.207) (BP4) and has no impact on splicing (SpliceAI = 0) (BP7). In summary, this variant is classified as a variant of uncertain significance for pulmonary arterial hypertension based on the

Met criteria codes

BP7			BP4 is met and the SpliceAI score (0) predicts no impact on splicing.
BP4			REVEL score not available. CADD = 1.4 is below the PH VCEP established threshold for benignity of <= 10.
PM2_Supporting			Maximum allele frequency is 0.00002 in gnomAD v2.1.1 controls and 0.00006 in gnomAD v4.1.

Not Met criteria codes

BA1			Variant has less than 5% allele frequency.
BS1			Allele frequency is not greater due to disorder.
PS4			Very low allele frequency and not reported in the literature.
PM1			No functional domain using UniProt and NextProt.

Curation History



 

 

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