

Variant: *NM_001204.7(BMPR2):c.1698T>A (p.Ile566=)*

Version: 1.0

CA64042886 [↗](#)

898486 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 328d0d54-f6de-427b-a63c-08ed41237655

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HGVS expressions

NM_001204.7(BMPR2):c.1698T>A (p.Ile566=)

NM_001204.7:c.1698T>A

NC_000002.12:g.202555363T>A

CM000664.2:g.202555363T>A

NC_000002.11:g.203420086T>A

CM000664.1:g.203420086T>A

NC_000002.10:g.203128331T>A

NG_009363.1:g.184037T>A

ENST00000374580.10:c.1698T>A

ENST00000638587.1:c.1629T>A

ENST00000374574.2:c.1586+2475T>A

ENST00000374580.8:c.1698T>A

NM_001204.6:c.1698T>A

Uncertain Significance

Met criteria codes **3**

BP7

BP4

PM2_Supporting

Not Met criteria codes **3**

PM1

BS1

BA1

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.1698T>A variant is a synonymous (silent) variant (p.Ile566=) that is not predicted by SpliceAI to impact splicing (BP4, BP7). This variant is absent from gnomAD v2.1.1 controls (PM2_supporting). Due to conflicting evidence, this variant is classified as a variant of uncertain significance for pulmonary arterial hypertension based on the ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP: BP4, BP7, PM2_supporting. (VCEP specifications version 1.1, 1/18/2024)

Met criteria codes

BP7			SpliceAI predicts no impact on splicing and the nucleotide is not highly conserved
BP4			Low CADD score (no REVEL score available as variant is synonymous)
PM2_Supporting			Variant allele is absent from controls dataset (gnomAD v2.1.1)

Not Met criteria codes

PM1			Located in a domain of undefined function
BS1			Allele frequency is less than 0.1% in controls (gnomAD v2.1.1)
BA1			Allele frequency is less than 1% in controls (gnomAD v2.1.1)

Curation History



 

 

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