

Variant: *NM_001204.7(BMPR2):c.218C>G (p.Ser73Ter)*

Version: 1.0

CA278072 [↗](#)

8797 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3f3b87be-41dc-4c8f-a0ce-53b1877cfd32

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HGVS expressions

NM_001204.7(BMPR2):c.218C>G (p.Ser73Ter)

NM_001204.7:c.218C>G

NC_000002.12:g.202464950C>G

CM000664.2:g.202464950C>G

NC_000002.11:g.203329673C>G

CM000664.1:g.203329673C>G

NC_000002.10:g.203037918C>G

NG_009363.1:g.93624C>G

ENST00000374580.10:c.218C>G

ENST00000638587.1:c.143C>G

ENST00000374574.2:c.218C>G

ENST00000374580.8:c.218C>G

ENST00000479069.1:n.125C>G

NM_001204.6:c.218C>G

Pathogenic

Met criteria codes **3**

PM2_Supporting

PVS1

PS4_Supporting

Not Met criteria codes **3**

BA1

PM1

BS1

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 c.218C>G, (p.Ser73Ter) (NM_001204.7) variant in BMPR2 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMID: 16429395). This variant has been reported in 2 pulmonary arterial hypertension probands (PS4_Supporting) (PMID: 26387786 ; PMID: 32581362). This variant is absent from gnomAD v2.1.1 controls (PM2_Supporting). In summary, this variant meets the criteria to be

classified as pathogenic for pulmonary arterial hypertension based on the ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP (specification version 11, 1/18/2024): PVS1, PS4_Supporting, PM2_Supporting.

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1			The c.218C>G, (p.Ser73Ter) (NM_001204.7) variant in BMPR2 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 2 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMID: 16429395).
PS4_Supporting			This variant has been reported in 2 probands meeting pulmonary arterial hypertension (PS4_Supporting;PMID: 26387786 ; PMID: 32581362).

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			This variant does not reside within a region of BMPR2 that is defined as a mutational hotspot or critical functional domain by the ClinGen Pulmonary Hypertension VCEP.
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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