

Variant: *NM_001204.7(BMPR2):c.86A>G (p.Asn29Ser)*

Version: 1.0

CA2061034 [↗](#)

333638 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: fbdadf39-ffbe-40f5-a79c-af267f12dcf5

Approved on: 2024/9/10

Published on: 2024/9/10

HGVS expressions

NM_001204.7(BMPR2):c.86A>G (p.Asn29Ser)

NM_001204.7:c.86A>G

NC_000002.12:g.202464818A>G

CM000664.2:g.202464818A>G

NC_000002.11:g.203329541A>G

CM000664.1:g.203329541A>G

NC_000002.10:g.203037786A>G

NG_009363.1:g.93492A>G

ENST00000374580.10:c.86A>G

ENST00000638587.1:c.11A>G

ENST00000374574.2:c.86A>G

ENST00000374580.8:c.86A>G

NM_001204.6:c.86A>G

Likely Benign

Met criteria codes **1**

BS1

Not Met criteria codes **2**

BP4

PP3

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 BMPR2 c.86A>G missense variant is predicted to cause a substitution of asparagine to serine at amino acid position 29 (p.Asn29Ser) in exon 2 within the signal peptide domain. The highest population minor allele frequency in gnomAD v2.1.1 controls is 0.006265 (v4.1.0 is 0.007668) in African/African American population, which is higher than the ClinGen Pulmonary Hypertension VCEP threshold $\geq 0.1\%$ for BS1, and therefore meets this criterion. The computational predictor REVEL gives a score of 0.497 indicating neither PP3 (≥ 0.75) nor BP4 (≤ 0.25) met. SpliceAI algorithm predicts no deleterious effect on acceptor or donor splice site. In summary, the variant meets the criteria

to be classified as variant of likely benign for pulmonary arterial hypertension based on the ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP: BS1 (VCEP specification version 1.1, 1/18/2024).

Met criteria codes

BS1			The highest population minor allele frequency in gnomAD v2.1.1 controls is 0.006265 (v4.1.0 is 0.007668) in African/African American population, which is higher than the ClinGen Pulmonary Hypertension VCEP threshold $\geq 0.1\%$ for BS1, and therefore meets this criterion.
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Not Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.497 indicating neither PP3 (≥ 0.75) nor BP4 (≤ 0.25) met. SpliceAI algorithm predicts no deleterious effect on acceptor or donor splice site.
PP3			The computational predictor REVEL score for the variant is 0.276, which does not meet PP3 (≥ 0.75) nor BP4 (≤ 0.25).

Curation History



 

 

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