

Variant: *NM_001204.7(BMPR2):c.2948G>A (p.Arg983Gln)*

Version: 1.0

CA2061617 [↗](#)

333652 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 45d8759d-7438-4066-a6cc-7193f6524fdb

Approved on: 2024/5/3

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HGVS expressions

NM_001204.7:c.2948G>A

NM_001204.7(BMPR2):c.2948G>A (p.Arg983Gln)

NC_000002.12:g.202559777G>A

CM000664.2:g.202559777G>A

NC_000002.11:g.203424500G>A

CM000664.1:g.203424500G>A

NC_000002.10:g.203132745G>A

NG_009363.1:g.188451G>A

ENST00000374580.10:c.2948G>A

ENST00000638587.1:c.2879G>A

ENST00000374574.2:c.*75G>A

ENST00000374580.8:c.2948G>A

NM_001204.6:c.2948G>A

Likely Benign

Met criteria codes **1**

BS1

Not Met criteria codes **8**

BA1

BP4

BS2

PP3

PM1

PM5

PS1

PM2

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.2948G>A variant is a missense variant predicted to cause substitution of arginine to glutamine at amino acid position 983 (p.Arg983Gln). The highest population minor allele frequency in gnomAD v2.1.1 controls is 0.0034 (8/2358) in the Ashkenazi Jew population, which is higher than the ClinGen PH VCEP threshold (>0.1%) for BS1, and therefore meets this criterion (BS1). The computational predictor REVEL gives a score of 0.4, which is neither above nor below the thresholds predicting a damaging or benign impact on BMPR2 function. In summary, this variant meets the criteria to be classified as likely benign for pulmonary arterial hypertension

based on the ACMG/AMP criteria applied, as specified by the ClinGen Pulmonary Hypertension VCEP: BS1. (VCEP specifications version 1.1, 1/18/2024)

Met criteria codes

BS1			GnomAD v2.1.1 control population was evaluated. Allele frequency in Ashkenazi Jews control population is 0.3% (8/2358). The frequency is greater than 0.1% cutoff for BS1 for PAH.
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Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			The computational predictor REVEL gives a score of 0.4, which is neither above nor below the thresholds predicting a damaging or benign impact on BMPR2 function. SpliceAI does not predict an effect on splicing.
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			The computational predictor REVEL gives a score of 0.4, which is neither above nor below the thresholds predicting a damaging or benign impact on BMPR2 function.
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.
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 

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