

Variant: *NM_001204.7(BMPR2):c.1766A>G (p.Tyr589Cys)*

Version: 1.0

CA2061448 [↗](#)

425966 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 6a60247f-8518-4f06-8c9d-6a6d73623484

Approved on: 2024/5/3

Published on: 2024/5/3

HGVS expressions

NM_001204.7(BMPR2):c.1766A>G (p.Tyr589Cys)

NM_001204.7:c.1766A>G

NC_000002.12:g.202555431A>G

CM000664.2:g.202555431A>G

NC_000002.11:g.203420154A>G

CM000664.1:g.203420154A>G

NC_000002.10:g.203128399A>G

NG_009363.1:g.184105A>G

ENST00000374580.10:c.1766A>G

ENST00000638587.1:c.1697A>G

ENST00000374574.2:c.1586+2543A>G

ENST00000374580.8:c.1766A>G

NM_001204.6:c.1766A>G

Likely Benign

Met criteria codes **1**

BS1

Not Met criteria codes **4**

PM2

PM1

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.1766A>G variant is a missense variant predicted to result in a tyrosine to cysteine substitution at amino acid 589 (p.Tyr589Cys). The minor allele frequency in gnomAD v2.1.1 controls at a frequency of 0.001511 (22/14,556 alleles) which is above the cut-off of 0.1% for PAH (BS1 met). The variant is located outside of the critical extracellular and kinase domains of BMPR2 (PM1 not met). The REVEL score is 0.577, not exceeding the threshold of ≥ 0.75 (PP3 not met) or ≤ 0.25 (BP4 not met). No functional data was available and SpliceAI does not predict an effect on splicing. In summary, the 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 specification version 1.1, 1/18/2024).			
Met criteria codes			
BS1			This variant is found in the Finnish population of gnomAD v2.1.1 controls at a frequency of 0.001511 (22/14,556 alleles) which is above the cut-off of 0.1% for PAH.
Not Met criteria codes			
PM2			This variant is found in the Finnish population of gnomAD v2.1.1 controls at a frequency of 0.001511 (22/14,556 alleles) which is above the cut-off of 0.01% for PAH.
PM1			The amino acid change that results from this variant is outside of the critical extracellular and kinase domains.
BP4			The REVEL score for this variant is 0.577 which is above the threshold of <=0.25 defined by the PH VCEP. SpliceAI does not predict an effect on splicing.
PP3			The REVEL score for this variant is 0.577 which is below the threshold of >=0.75 defined by the PH VCEP and SpliceAI predicts no effect on splicing (score = 0).

Curation History



 

 

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.