

Variant: *NM_001204.7(BMPR2):c.545G>A (p.Gly182Asp)*

Version: 2.0

CA119936 [↗](#)

8813 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 370663a6-33e3-49fb-b66f-293d99e1c5c7

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HGVS expressions

NM_001204.7:c.545G>A

NM_001204.7(BMPR2):c.545G>A (p.Gly182Asp)

NC_000002.12:g.202514903G>A

CM000664.2:g.202514903G>A

NC_000002.11:g.203379626G>A

CM000664.1:g.203379626G>A

NC_000002.10:g.203087871G>A

NG_009363.1:g.143577G>A

ENST00000374580.10:c.545G>A

ENST00000638587.1:c.476G>A

ENST00000374574.2:c.545G>A

ENST00000374580.8:c.545G>A

NM_001204.6:c.545G>A

Uncertain Significance

Met criteria codes **1**

BS3

Not Met criteria codes **5**

PS1

PP3

PM5

PM2

BS1

Evidence Links **1**

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 2.0.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Pulmonary Hypertension VCEP

The NM_0001204.7(BMPR2) c.545G>A variant is a missense variant predicted to cause substitution of glycine by aspartic acid at amino acid 182 (p.Gly182Asp). The highest subpopulation minor allele frequency in gnomAD v2.1.1 (controls) is 0.0003507 in the European (non-Finnish) population, which is higher than the ClinGen PH VCEP threshold (<0.01%) for PM2 but lower than the threshold (>0.1%) for BS1. A similar AF in gnomAD v4.1 (non-Finnish Europeans, 388/1179912) confirms the non-application of PM2 (PM2 not met). Computational prediction tool scores (REVEL 0.8069, AlphaMissense 0.4252, CADD 16.92) did not yield two out of three of these above the PH VCEP

thresholds (PP3 not met). Luciferase assay data indicated that variant transcriptional activity was comparable to wild-type, indicating no deleterious effect (BS3; PMID: 18321866). 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: no criteria scored. (VCEP specifications version 2.0, 1/30/2026)

Met criteria codes

BS3			Cells transfected with the p.G182D variant have no damaging effect on transcriptional activity by luciferase assay (PMID: 18321866)
			Transcriptional activity of the p.G182D variant is comparable to wild-type using a luciferase assay PubMed:18321866 

Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			Computational prediction tool scores (REVEL 0.8069, AlphaMissense 0.4252, CADD 16.92) did not yield two out of three of these above the PH VCEP thresholds.
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			Allele frequency in the European population (gnomAD v2.1.1 controls) is greater than 0.01% (MAF: 15/42768 = 0.0003507). A similar AF in gnomAD v4.1 (non-Finnish Europeans, 388/1179912) confirms the non-application of PM2..
BS1			Allele frequency is less than 0.1% in gnomAD v2.1.1 controls

Curation History



 

 

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