

Variant: *NM_001204.7(BMPR2):c.1126G>T (p.Glu376Ter)*

Version: 1.0

CA350341605 [↗](#)

425876 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0727e26f-4fdd-43b6-84dc-1ca98dc8dc80

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HGVS expressions

NM_001204.7(BMPR2):c.1126G>T (p.Glu376Ter)

NM_001204.7:c.1126G>T

NC_000002.12:g.202530952G>T

CM000664.2:g.202530952G>T

NC_000002.11:g.203395675G>T

CM000664.1:g.203395675G>T

NC_000002.10:g.203103920G>T

NG_009363.1:g.159626G>T

ENST00000374580.10:c.1126G>T

ENST00000638587.1:c.1057G>T

ENST00000374574.2:c.1126G>T

ENST00000374580.8:c.1126G>T

NM_001204.6:c.1126G>T

Pathogenic

Met criteria codes **3**

PM2

PS4_Supporting

PVS1

Not Met criteria codes **2**

BA1

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

BMPR2 c.1126G>T (p.Glu376Ter) variant is predicted to cause premature stop at residue 376, in exon 8, and lead to nonsense mediated decay. The variant is absent from gnomAD v2.1.1 and v4.1 (PM2_supporting met) and was identified in two unrelated idiopathic PAH probands (PMID: 26387786 and PMID: 31727138) (PS4_supporting met). In summary, the 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: PVS1, PM2_supporting, PS4_supporting (VCEP specification version 1.1, 1/18/2024).

Met criteria codes

PM2		✓	The variant is absent from gnomAD v2.1.1 and v4.1.
PS4_Supporting		✓	The variant was identified in two unrelated idiopathic pulmonary arterial hypertension patients (PMID: 26387786 and PMID: 31727138).
PVS1		✓	The variant is a nonsense variant predicted to cause premature stop at residue 376, in exon 8, leading to nonsense mediated decay.

Not Met criteria codes

BA1		✗	The variant is absent from gnomAD v2.1.1 and v4.1.
BS1		✗	The variant is absent from gnomAD v2.1.1 and v4.1.

Curation History



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