

Variant: *NM_024675.3(PALB2):c.135G>A (p.Lys45=)*

Version: 2.0

CA7963834 [↗](#)

230997 (ClinVar) [↗](#)

Gene: PALB2 ([HGNC:79728](#))

Condition: PALB2-related cancer predisposition ([MONDO:0700272](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 9513d8df-0b94-4551-975f-a4691e9f1520

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HGVS expressions

NM_024675.3:c.135G>A

NM_024675.3(PALB2):c.135G>A (p.Lys45=)

NC_000016.10:g.23637926C>T

CM000678.2:g.23637926C>T

NC_000016.9:g.23649247C>T

CM000678.1:g.23649247C>T

NC_000016.8:g.23556748C>T

NG_007406.1:g.8432G>A

ENST00000561514.3:c.141G>A

ENST00000565038.2:c.135G>A

ENST00000566069.6:c.135G>A

ENST00000697377.2:c.141G>A

ENST00000697379.2:c.141G>A

ENST00000561514.2:c.-751G>A

ENST00000697374.1:c.-751G>A

ENST00000697375.1:n.1482G>A

ENST00000697376.1:c.-751G>A

ENST00000697377.1:c.-751G>A

ENST00000697378.1:n.655G>A

ENST00000697379.1:c.-751G>A

ENST00000697382.1:c.-751G>A

ENST00000697383.1:c.48+3184G>A

ENST00000697384.1:n.289G>A

ENST00000261584.9:c.135G>A

ENST00000261584.8:c.135G>A

ENST00000561514.1:c.141G>A

ENST00000565038.1:c.10G>A

ENST00000567003.1:n.413G>A

ENST00000568219.5:c.-751G>A

NM_024675.4:c.135G>A

Likely Benign

Met criteria codes 2

BP7BP4

Not Met criteria codes 2

PM2BS1

Evidence Links 0

Expert Panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

Criteria Specification Information

Criteria Specification: ClinGen Hereditary Breast, Ovarian and Pancreatic Cancer Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PALB2 Version 1.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.135G>A (p.Lys45=) variant is a synonymous (silent) variant that is not predicted by MaxEntScan or SpliceAI to impact splicing. This variant has a minor allele frequency in gnomAD v2.1.1 of 0.000008791 in the European non-Finnish population (PM2_Supporting, BS1, and BA1 are not met). In summary, this variant meets criteria to be classified as likely benign for autosomal dominant hereditary breast and pancreatic cancer and autosomal recessive FANCN based on the ACMG/AMP criteria applied as specified by the HBOP VCEP. (BP4, BP7)

Met criteria codes

BP7

The c.135G>A (p.Lys45=) variant is a synonymous (silent) variant that is not predicted by MaxEntScan or SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown (BP4, BP7).

BP4

In silico predictors (MaxEntScan/SpliceAI) are in agreement that this variant is benign (BP4), can be used with BP7 per HBOP VCEP.

Not Met criteria codes

PM2

Minor allele frequency in gnomAD v2.1.1 of 0.00001 in the European non-Finnish population

BS1

Minor allele frequency in gnomAD v2.1.1 of 0.00001 in the European non-Finnish population

Curation History

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