

Variant: *NM_024675.4(PALB2):c.1748T>G (p.Leu583Trp)*

Version: 2.0

CA166979 [↗](#)

141972 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 784d2c84-8207-4f86-908e-4646f2ce3c66

Approved on: 2023/4/5

Published on: 2025/9/16

HGVS expressions

NM_024675.4:c.1748T>G

NM_024675.4(PALB2):c.1748T>G (p.Leu583Trp)

NC_000016.10:g.23630406A>C

CM000678.2:g.23630406A>C

NC_000016.9:g.23641727A>C

CM000678.1:g.23641727A>C

NC_000016.8:g.23549228A>C

NG_007406.1:g.15952T>G

ENST00000561514.3:c.1754T>G

ENST00000565038.2:c.212-1131T>G

ENST00000566069.6:c.1748T>G

ENST00000697377.2:c.1754T>G

ENST00000697379.2:c.1754T>G

ENST00000561514.2:c.863T>G

ENST00000697374.1:c.863T>G

ENST00000697375.1:n.3095T>G

ENST00000697376.1:c.863T>G

ENST00000697377.1:c.863T>G

ENST00000697378.1:n.2268T>G

ENST00000697379.1:c.863T>G

ENST00000697380.1:n.676T>G

ENST00000697381.1:n.443T>G

ENST00000697382.1:c.863T>G

ENST00000697383.1:c.49-1131T>G

ENST00000697384.1:n.1902T>G

ENST00000261584.9:c.1748T>G

ENST00000261584.8:c.1748T>G

ENST00000565038.1:c.87-1131T>G

ENST00000568219.5:c.863T>G

NM_024675.3:c.1748T>G

Uncertain Significance

Met criteria codes 1

BP1

Not Met criteria codes 3

BA1BS1PM2

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.1748T>G (p.Leu583Trp) variant in PALB2 is a missense variant predicted to cause a substitution of leucine by tryptophan at amino acid 583 (p.Leu583Trp). This variant has a minor allele frequency in gnomAD v2.1.1 of 0.000044 in the non-Finnish European population (PM2_Supporting, BS1, and BA1 are not met). PALB2, in which the variant was identified, is defined by the HBOP VCEP as a gene for which primarily truncating variants are known to cause disease. In summary, this variant meets the criteria to be classified as a variant of uncertain significance 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. (BP1)

Met criteria codes

BP1

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

BA1

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS1

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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