

Variant: *NM\_024675.3(PALB2):c.532del (p.Glu178fs)*

Version: 2.0

[CA891843628](#) 

[580962 \(ClinVar\)](#) 

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UID:** dbcda545-7d56-41f0-9e04-3e0cb47d842b

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### *HGVS expressions*

**NM\_024675.3:c.532del**

**NM\_024675.3:c.532delG**

NM\_024675.3(PALB2):c.532del (p.Glu178fs)

NC\_000016.10:g.23636015del

CM000678.2:g.23636015del

NC\_000016.9:g.23647336del

CM000678.1:g.23647336del

NC\_000016.8:g.23554837del

NG\_007406.1:g.10344del

ENST00000561514.3:c.538del

ENST00000565038.2:c.211+1836del

ENST00000566069.6:c.532del

ENST00000697377.2:c.538del

ENST00000697379.2:c.538del

ENST00000561514.2:c.-354del

ENST00000697374.1:c.-354del

ENST00000697375.1:n.1879del

ENST00000697376.1:c.-354del

ENST00000697377.1:c.-354del

ENST00000697378.1:n.1052del

ENST00000697379.1:c.-354del

ENST00000697382.1:c.-354del

ENST00000697383.1:c.48+5096del

ENST00000697384.1:n.686del

ENST00000261584.9:c.532del

ENST00000261584.8:c.532del

ENST00000565038.1:c.86+1836del

ENST00000568219.5:c.-354del

NM\_024675.4:c.532del

**Pathogenic**

**Met criteria codes** **3**

**PM5\_Supporting** **PVS1**

**PM2\_Supporting**

**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.532del (p.Glu178fs) variant in PALB2 is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 4 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism. This alteration results in a termination codon upstream of the most C-terminal pathogenic alteration (PALB2 p.Tyr1183\*), as classified by the HBOP VCEP, and is expected to be more deleterious. This variant is absent in gnomAD v2.1.1. In summary, this variant meets the criteria to be classified as pathogenic 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. (PVS1, PM2\_Supporting, PM5\_Supporting)

#### Met criteria codes

<b>PM5_Supporting</b>			This alteration results in a termination codon upstream of the most C-terminal pathogenic alteration (PALB2 p.Tyr1183Ter), as classified by the ClinGen Hereditary Breast, Ovarian, and Pancreatic Cancer VCEP, and is expected to be more deleterious (PM5_Supporting).
<b>PVS1</b>			The c.532del (p.Glu178fs) (NM_024675 .3) variant in PALB2 is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 4 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
<b>PM2_Supporting</b>			Variant is absent in the GnomAD v2.1.1 (PM2_Supporting)

Curation History [↗](#)



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