The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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
UUID: dbcda545-7d56-41f0-9e04-3e0cb47d842b
Approved on: 2023/4/5
Published on: 2025/9/16

HGVS expressions

NM_024675.3:c.532delG
NM_024675.3:c.532del
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

PVS1 PM2_Supporting PM5_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

PVS1	✔	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).
PM2_Supporting	✔	Variant is absent in the GnomAD v2.1.1 (PM2_Supporting)
PM5_Supporting	✔	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).

Curation History

Showing 1 to 2 of 2 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.