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_024675.4:c.2787_2788dup

Version: 2.0

CA915941070

1453402 (ClinVar)

Gene: PALB2 (HGNC:79728)
Condition: PALB2-related cancer predisposition (MONDO:0700272)
Inheritance Mode: Autosomal dominant inheritance
UUID: 5e667e74-d7fc-4604-bac1-2edcf9f0c2df
Approved on: 2023/4/5
Published on: 2025/9/16

HGVS expressions

NM_024675.4:c.2787_2788dup
NC_000016.10:g.23624057_23624058dup
CM000678.2:g.23624057_23624058dup
NC_000016.9:g.23635378_23635379dup
CM000678.1:g.23635378_23635379dup
NC_000016.8:g.23542879_23542880dup
NG_007406.1:g.22302_22303dup
ENST00000561514.3:c.2793_2794dup
ENST00000565038.2:c.*268_*269dup
ENST00000566069.6:c.2787_2788dup
ENST00000697377.2:c.2631_2632dup
ENST00000697379.2:c.2793_2794dup
ENST00000561514.2:c.1902_1903dup
ENST00000697374.1:c.1902_1903dup
ENST00000697375.1:n.4134_4135dup
ENST00000697376.1:c.1902_1903dup
ENST00000697377.1:c.1740_1741dup
ENST00000697378.1:n.3307_3308dup
ENST00000697379.1:c.1902_1903dup
ENST00000697380.1:n.2079_2080dup
ENST00000697381.1:n.1482_1483dup
ENST00000697382.1:c.1902_1903dup
ENST00000697383.1:c.321_322dup
ENST00000261584.9:c.2787_2788dup
ENST00000261584.8:c.2787_2788dup
ENST00000568219.5:c.1902_1903dup
NM_024675.3:c.2787_2788dup

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting 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.2787_2788dup (p.Asn930Ilefs*6) variant in PALB2 is a frameshift variant predicted to cause a premature stop codon in biologically-relevant exon 8 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism. This variant is absent in gnomAD v2.1.1. This alteration results in a termination codon upstream of the most C-terminus pathogenic alteration (PALB2 p.Tyr1183*) as classified by the HBOP VCEP, and is expected to be more deleterious. In summary, this variant meets 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 p.N930Ifs*6 variant is predicted to result in a premature stop codon that leads to truncated or absent protein (PVS1)
PM5_Supporting	✔	This alteration results in a termination codon upstream of the most C-terminus pathogenic alteration (PALB2, p.Tyr1183*) as classified by the ClinGen HBOP Variant Curation Expert Panel, and is expected to be more deleterious (PM5_Supporting).
PM2_Supporting	✔	Variant is absent in the GnomAD v2.1.1 (PM2_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.