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Variant: NM_000059.4(BRCA2):c.8243G>T (p.Gly2748Val)

Version: 1.0

CA387749910

1171382 (ClinVar)

Gene: BRCA2 (HGNC:675)
Condition: BRCA2-related cancer predisposition (MONDO:0700269)
Inheritance Mode: Autosomal dominant inheritance
UUID: 4020fc9a-ccd5-4b9c-b352-5ce05568e0da
Approved on: 2026/7/15
Published on: 2026/7/15

HGVS expressions

NM_000059.4(BRCA2):c.8243G>T (p.Gly2748Val)
NM_000059.4:c.8243G>T
NC_000013.11:g.32363445G>T
CM000675.2:g.32363445G>T
NC_000013.10:g.32937582G>T
CM000675.1:g.32937582G>T
NC_000013.9:g.31835582G>T
NG_012772.3:g.52966G>T
ENST00000470094.2:c.8243G>T
ENST00000528762.2:c.8243G>T
ENST00000530893.7:c.7874G>T
ENST00000665585.2:c.8243G>T
ENST00000666593.2:c.8243G>T
ENST00000700202.2:c.8243G>T
ENST00000700202.1:c.710G>T
ENST00000380152.8:c.8243G>T
ENST00000544455.6:c.8243G>T
ENST00000614259.2:c.8251G>T
ENST00000665585.1:c.808G>T
ENST00000680887.1:c.8243G>T
ENST00000380152.7:c.8243G>T
ENST00000544455.5:c.8243G>T
NM_000059.3:c.8243G>T

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PS3 PP3

Evidence Links 0

Expert Panel

ENIGMA BRCA1 and BRCA2 VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen ENIGMA BRCA1 and BRCA2 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for BRCA2 Version 1.1.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The c.8243G>T variant in BRCA2 is a missense variant predicted to cause substitution of glycine by valine at amino acid 2748 (p.(p.Gly2748Val)). This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met). This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.45, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.00 predicts no impact on splicing (score threshold ≤0.1) (PP3). Reported by two calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs: 39779857, 39779848) (PS3 met). In summary, this variant meets the criteria to be classified as a Likely pathogenic variant for BRCA2-related cancer predisposition based on the ACMG/AMP criteria applied as specified by the ENIGMA BRCA1/2 VCEP (PM2_Supporting, PP3, PS3).

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met).
PS3	✔	Reported by two calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs: 39779857, 39779848) (PS3 met).
PP3	✔	This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.45, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.00 predicts no impact on splicing (score threshold ≤0.1) (PP3 met).

Curation History

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