

Variant: *NM_000059.4(BRCA2):c.7955T>G (p.Val2652Gly)*

Version: 1.0

CA387747262 [↗](#)

489784 (ClinVar) [↗](#)

Gene: BRCA2 ([HGNC:675](#))

Condition: BRCA2-related cancer predisposition ([MONDO:0700269](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 11df9d69-ba86-4bb3-8a00-dd5d5f949030

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HGVS expressions

NM_000059.4(BRCA2):c.7955T>G (p.Val2652Gly)

NM_000059.4:c.7955T>G

NC_000013.11:g.32362672T>G

CM000675.2:g.32362672T>G

NC_000013.10:g.32936809T>G

CM000675.1:g.32936809T>G

NC_000013.9:g.31834809T>G

NG_012772.3:g.52193T>G

ENST00000470094.2:c.7955T>G

ENST00000528762.2:c.7955T>G

ENST00000530893.7:c.7586T>G

ENST00000665585.2:c.7955T>G

ENST00000666593.2:c.7955T>G

ENST00000700202.2:c.7955T>G

ENST00000700202.1:c.422T>G

ENST00000380152.8:c.7955T>G

ENST00000544455.6:c.7955T>G

ENST00000614259.2:c.7963T>G

ENST00000665585.1:c.520T>G

ENST00000680887.1:c.7955T>G

ENST00000380152.7:c.7955T>G

ENST00000544455.5:c.7955T>G

NM_000059.3:c.7955T>G

Likely Pathogenic

Met criteria codes **3**

PS3

PP3

PM2_Supporting

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**

ENIGMA BRCA1 and BRCA2 VCEP

The c.7955T>G variant in BRCA2 is a missense variant predicted to cause substitution of Valine by Glycine at amino acid 2652 (p. (Val2652Gly)). This variant is absent from gnomAD v2.1 (exomes only, non-cancer subset, read depth ≥25) and gnomAD v3.1 (non-cancer subset, read depth ≥25) (PM2_Supporting met). Reported by five calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 32444794, 35190686, 39779857, 39779848) (PS3 met). This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.42, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (PP3 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, PS3, PP3).

Met criteria codes

PS3			Reported by five calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 32444794, 35190686, 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 0.42, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (PP3 met).
PM2_Supporting			This variant is absent from gnomAD v2.1 (exomes only, non-cancer subset, read depth ≥25) and gnomAD v3.1 (non-cancer subset, read depth ≥25) (PM2_Supporting met).

Curation History



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