

Variant: *NM_000059.4(BRCA2):c.7855T>C (p.Trp2619Arg)*

Version: 1.0

CA387747058 [↗](#)

827289 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9a3cdabb-780e-46c8-9be4-c236c211019e

Approved on: 2026/7/15

Published on: 2026/7/15

HGVS expressions

NM_000059.4:c.7855T>C

NM_000059.4(BRCA2):c.7855T>C (p.Trp2619Arg)

NC_000013.11:g.32362572T>C

CM000675.2:g.32362572T>C

NC_000013.10:g.32936709T>C

CM000675.1:g.32936709T>C

NC_000013.9:g.31834709T>C

NG_012772.3:g.52093T>C

ENST00000470094.2:c.7855T>C

ENST00000528762.2:c.7855T>C

ENST00000530893.7:c.7486T>C

ENST00000665585.2:c.7855T>C

ENST00000666593.2:c.7855T>C

ENST00000700202.2:c.7855T>C

ENST00000700202.1:c.322T>C

ENST00000380152.8:c.7855T>C

ENST00000544455.6:c.7855T>C

ENST00000614259.2:c.7863T>C

ENST00000665585.1:c.420T>C

ENST00000680887.1:c.7855T>C

ENST00000380152.7:c.7855T>C

ENST00000544455.5:c.7855T>C

ENST00000614259.1:n.7863T>C

NM_000059.3:c.7855T>C

Likely Pathogenic

Met criteria codes **3**

PS3 PP3 PM2_Supporting

Not Met criteria codes **6**

PS1 PP1 PP4 BS4 PM5
BP5

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 1.2.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

ENIGMA BRCA1 and BRCA2 VCEP

The variant c.7855T>C in BRCA2 is a missense variant predicted to cause substitution of Tryptophan by Arginine at amino acid 2619 (p.Trp2619Arg). 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 0.49, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.08 predicts no impact on splicing (score threshold ≤0.1) (PP3 met). Reported by three calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 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

PS3			Reported by three calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 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 0.49, above the recommended threshold of 0.30 for prediction of impact on BRCA2 function via protein change. A SpliceAI score of 0.08 predicts no impact on splicing (score threshold ≤0.1) (PP3 met).
PM2_Supporting			This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met).

Not Met criteria codes

PS1			(BRCA2):c.7857G>C (p.Trp2619Cys) is classified as LP by VCEP; however, there are no p.Trp2619Arg variants.
PP1			No LR/segregations studies for this variant.
PP4			No LR/segregations studies for this variant.
BS4			No LR/segregations studies for this variant.
PM5			(BRCA2):c.7857G>C (p.Trp2619Cys) is classified as LP by VCEP; however, this code does not capture different missense substitutions.
BP5			No LR/segregations studies for this variant.











Showing 1 to 1 of 1 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.