

Variant: *NM_000059.4(BRCA2):c.7819A>C (p.Thr2607Pro)*

Version: 1.0

CA025298 [↗](#)

52422 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 2147d84b-0db5-4482-a760-0b62b024c829

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HGVS expressions

NM_000059.4:c.7819A>C

NM_000059.4(BRCA2):c.7819A>C (p.Thr2607Pro)

NC_000013.11:g.32362536A>C

CM000675.2:g.32362536A>C

NC_000013.10:g.32936673A>C

CM000675.1:g.32936673A>C

NC_000013.9:g.31834673A>C

NG_012772.3:g.52057A>C

ENST00000470094.2:c.7819A>C

ENST00000528762.2:c.7819A>C

ENST00000530893.7:c.7450A>C

ENST00000665585.2:c.7819A>C

ENST00000666593.2:c.7819A>C

ENST00000700202.2:c.7819A>C

ENST00000700202.1:c.286A>C

ENST00000380152.8:c.7819A>C

ENST00000544455.6:c.7819A>C

ENST00000614259.2:c.7827A>C

ENST00000665585.1:c.384A>C

ENST00000680887.1:c.7819A>C

ENST00000380152.7:c.7819A>C

ENST00000544455.5:c.7819A>C

ENST00000614259.1:n.7827A>C

NM_000059.3:c.7819A>C

Likely Pathogenic

Met criteria codes 3

PP4_Moderate

PS3

PM2_Supporting

Not Met criteria codes 4

BP7

BP4

BP1

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 1.2.0

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

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

ENIGMA BRCA1 and BRCA2 VCEP

The c.7819A>C variant in BRCA2 is a missense variant predicted to cause substitution of Threonine by Proline at amino acid 2607 (p. (Thr2607Pro)). This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met). Reported by three calibrated studies to exhibit functional effect similar to pathogenic control variants (PMIDs: 38417439, 39779857, 39779848) and reported by one calibrated study to show an impact of varying degree in different assays (PMID: 32444794) (PS3 met). This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.23, indicating impact on BRCA2 function via protein change is unclear (score range 0.18-0.30). A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (no bioinformatic code is applied). Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 11.9 (based on Co-occurrence LR=1.03; Family History LR=11.62), within the thresholds for moderate evidence towards pathogenicity (LR >4.3 & ≤18.7) (PP4_Moderate met; Internal lab contributors). 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, PP4_Moderate, PS3).

Met criteria codes

PP4_Moderate			Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 11.9 (based on Co-occurrence LR=1.03; Family History LR=11.62), within the thresholds for moderate evidence towards pathogenicity (LR >4.3 & ≤18.7) (PP4_Moderate met; Internal lab contributors).
PS3			Reported by three calibrated studies to exhibit functional effect similar to pathogenic control variants (PMIDs: 38417439, 39779857, 39779848) and reported by one calibrated study to show an impact of varying degree in different assays (PMID: 32444794) (PS3 met).
PM2_Supporting			This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met).

Not Met criteria codes

BP7			This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.23, indicating impact on BRCA2 function via protein change is unclear (score range 0.18-0.30). A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (no bioinformatic code is applied).
BP4			This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.23, indicating impact on BRCA2 function via protein change is unclear (score range 0.18-0.30). A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (no bioinformatic code is applied).
BP1			This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.23, indicating impact on BRCA2 function via protein change is unclear (score range 0.18-0.30). A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (no bioinformatic code is applied).
PP3			This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.23, indicating impact on BRCA2 function via protein change is unclear (score range 0.18-0.30). A SpliceAI score of 0.01 predicts no impact on splicing (score threshold <0.10) (no bioinformatic code is applied).

Curation History 

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