

Variant: *NM_000059.4(BRCA2):c.9372C>A (p.Asn3124Lys)*

Version: 1.0

CA387761213 [↗](#)

462524 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0d707dd7-0bb1-4186-8bc4-eb9044fcc52e

Approved on: 2026/1/5

Published on: 2026/1/5

HGVS expressions

NM_000059.4(BRCA2):c.9372C>A (p.Asn3124Lys)

NM_000059.4:c.9372C>A

NC_000013.11:g.32394804C>A

CM000675.2:g.32394804C>A

NC_000013.10:g.32968941C>A

CM000675.1:g.32968941C>A

NC_000013.9:g.31866941C>A

NG_012772.3:g.84325C>A

ENST00000470094.2:c.9372C>A

ENST00000528762.2:c.*739C>A

ENST00000530893.7:c.9003C>A

ENST00000665585.2:c.*934C>A

ENST00000666593.2:c.*217C>A

ENST00000700202.2:c.9321C>A

ENST00000700202.1:c.1788C>A

ENST00000700203.1:n.1499C>A

ENST00000380152.8:c.9372C>A

ENST00000544455.6:c.9372C>A

ENST00000614259.2:c.9380C>A

ENST00000665585.1:c.2250C>A

ENST00000666593.1:c.394C>A

ENST00000680887.1:c.9372C>A

ENST00000380152.7:c.9372C>A

ENST00000470094.1:c.329C>A

ENST00000544455.5:c.9372C>A

NM_000059.3:c.9372C>A

Uncertain Significance

Met criteria codes **3**

PM2_Supporting BP4 PS3

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

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The c.9372C>A variant in BRCA2 is a missense variant predicted to cause substitution of Asparagine by Lysine at amino acid 3124 (p. (Asn3124Lys)). 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 two calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 39779857) (PS3 met). This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.0006, which is below the recommended threshold of 0.18 for predicting no impact on BRCA2 via protein change. A SpliceAI score of 0 predicts no impact on splicing (score threshold < 0.10) (BP4 met). In summary, this variant meets the criteria to be classified as a Variant of uncertain significance for BRCA2-related cancer predisposition based on the ACMG/AMP criteria applied as specified by the ENIGMA BRCA1/2 VCEP (PM2_Supporting, PS3, BP4).

Met criteria codes

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).
BP4			This BRCA2 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.0006, which is below the recommended threshold of 0.18 for predicting no impact on BRCA2 via protein change. A SpliceAI score of 0 predicts no impact on splicing (score threshold < 0.10) (BP4 met).
PS3			Reported by two calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:38417439, 39779857) (PS3 met).

Curation History [↗](#)



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