

Variant: *NM_007294.4(BRCA1):c.286G>A (p.Asp96Asn)*

Version: 1.0

CA001871 [↗](#)

54704 (ClinVar) [↗](#)

Gene: BRCA1 ([HGNC:672](#))

Condition: BRCA1-related cancer predisposition ([MONDO:0700268](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: ca89c16c-f8e4-46d9-89ba-b95ad2c5a405

Approved on: 2026/1/5

Published on: 2026/1/5

HGVS expressions

NM_007294.4:c.286G>A

NM_007294.4(BRCA1):c.286G>A (p.Asp96Asn)

NC_000017.11:g.43104883C>T

CM000679.2:g.43104883C>T

NC_000017.10:g.41256900C>T

CM000679.1:g.41256900C>T

NC_000017.9:g.38510426C>T

NG_005905.2:g.113101G>A

ENST00000354071.8:n.350G>A

ENST00000461574.2:c.286G>A

ENST00000470026.6:c.286G>A

ENST00000473961.6:c.286G>A

ENST00000476777.6:c.286G>A

ENST00000477152.6:c.208G>A

ENST00000478531.6:c.286G>A

ENST00000489037.2:c.208G>A

ENST00000493919.6:c.145G>A

ENST00000494123.6:c.286G>A

ENST00000497488.2:c.-218-10023G>A

ENST00000618469.2:c.286G>A

ENST00000634433.2:c.286G>A

ENST00000644379.2:c.286G>A

ENST00000644555.2:c.145G>A

ENST00000652672.2:c.145G>A

ENST00000484087.6:c.286G>A

ENST00000700083.1:n.1257G>A

ENST00000700182.1:c.208G>A

ENST00000700183.1:c.*200G>A

ENST00000700184.1:n.529G>A

ENST00000357654.9:c.286G>A

ENST00000471181.7:c.286G>A

ENST00000642945.1:c.*160G>A

ENST00000644555.1:c.145G>A

ENST00000652672.1:c.145G>A

ENST00000352993.7:c.286G>A

ENST00000354071.7:c.286G>A

ENST00000357654.7:c.286G>A

ENST00000461221.5:c.*72G>A
ENST00000468300.5:c.286G>A
ENST00000470026.5:c.286G>A
ENST00000471181.6:c.286G>A
ENST00000473961.5:c.9G>A
ENST00000476777.5:c.286G>A
ENST00000477152.5:c.208G>A
ENST00000478531.5:c.286G>A
ENST00000484087.5:c.34G>A
ENST00000487825.5:c.34G>A
ENST00000489037.1:c.208G>A
ENST00000491747.6:c.286G>A
ENST00000492859.5:c.*222G>A
ENST00000493795.5:c.145G>A
ENST00000493919.5:c.145G>A
ENST00000494123.5:c.286G>A
ENST00000497488.1:c.-218-10023G>A
ENST00000586385.5:c.4+20299G>A
ENST00000591534.5:c.-44+20388G>A
ENST00000591849.5:c.-99+20388G>A
ENST00000634433.1:c.286G>A
NM_007294.3:c.286G>A
NM_007297.3:c.145G>A
NM_007298.3:c.286G>A
NM_007299.3:c.286G>A
NM_007300.3:c.286G>A
NR_027676.1:n.425G>A
NM_007297.4:c.145G>A
NM_007299.4:c.286G>A
NM_007300.4:c.286G>A
NR_027676.2:n.466G>A

Likely Pathogenic

Met criteria codes 3

PP4_Moderate

PS3

PM2_Supporting

Not Met criteria codes 4

BP4

BP1

BP7

PP3

Evidence Links 0

Expert Panel

ENIGMA BRCA1 and BRCA2 VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The c.286G>A variant in BRCA1 is a missense variant predicted to cause substitution of Aspartic Acid by Asparagine at amino acid 96 (p.(Asp96Asn)). 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). This BRCA1 missense variant is within a key functional domain and the computational

predictor BayesDel (noAF) gives a score of 0.077, below the recommended threshold of 0.15 for no predicted impact on BRCA1 function via protein change. However, the SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score range 0.10-0.20) (no bioinformatic code is applied). Reported by two calibrated studies to exhibit protein function similar to pathogenic control variants (PMID:35659930, Findlay internal data) (PS3 met). mRNA experimental analysis indicates no impact on splicing (PMID: 32123317) but is not applied as strong evidence against pathogenicity since missense impact has not been excluded (BP7_Strong (RNA) not met). Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 9.53 (based on Family History LR=9.53), within the thresholds for moderate evidence towards pathogenicity (LR >4.3 & ≤18.7) (PP4_Moderate met; PMID: 31853058). In summary, this variant meets the criteria to be classified as a Likely pathogenic variant for BRCA1-related cancer predisposition based on the ACMG/AMP criteria applied as specified by the ENIGMA BRCA1/2 VCEP (PM2_Supporting, PS3, PP4_Moderate).

Met criteria codes

PP4_Moderate			Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 9.53 (based on Family History LR=9.53), within the thresholds for moderate evidence towards pathogenicity (LR >4.3 & ≤18.7) (PP4_Moderate met; PMID: 31853058).
PS3			Reported by two calibrated studies to exhibit protein function similar to pathogenic control variants (PMID:35659930, Findlay internal data) (PS3 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).

Not Met criteria codes

BP4			This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.077, below the recommended threshold of 0.15 for no predicted impact on BRCA1 function via protein change. However, the SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score range 0.10-0.20) (no bioinformatic code is applied).
BP1			This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.077, below the recommended threshold of 0.15 for no predicted impact on BRCA1 function via protein change. However, the SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score range 0.10-0.20) (no bioinformatic code is applied).
BP7			mRNA experimental analysis indicates no impact on splicing (PMID: 32123317) but is not applied as strong evidence against pathogenicity since missense impact has not been excluded (BP7_Strong (RNA) not met).
PP3			This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.077, below the recommended threshold of 0.15 for no predicted impact on BRCA1 function via protein change. However, the SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score range 0.10-0.20) (no bioinformatic code is applied).

▼

▼

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.