The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_007294.4(BRCA1):c.56A>C (p.Gln19Pro)

Version: 1.0

CA10602046

867727 (ClinVar)

Gene: BRCA1 (HGNC:672)
Condition: BRCA1-related cancer predisposition (MONDO:0700268)
Inheritance Mode: Autosomal dominant inheritance
UUID: 7febf7e8-13a6-42fa-8c0c-1302a42cd43b
Approved on: 2026/8/28
Published on: 2026/8/27

HGVS expressions

NM_007294.4(BRCA1):c.56A>C (p.Gln19Pro)
NM_007294.4:c.56A>C
NC_000017.11:g.43124041T>G
CM000679.2:g.43124041T>G
NC_000017.10:g.41276058T>G
CM000679.1:g.41276058T>G
NC_000017.9:g.38529584T>G
NG_005905.2:g.93943A>C
ENST00000354071.8:n.120A>C
ENST00000461574.2:c.56A>C
ENST00000470026.6:c.56A>C
ENST00000473961.6:c.56A>C
ENST00000476777.6:c.56A>C
ENST00000477152.6:c.56A>C
ENST00000478531.6:c.56A>C
ENST00000489037.2:c.56A>C
ENST00000493919.6:c.-32A>C
ENST00000494123.6:c.56A>C
ENST00000497488.2:c.-219+1230A>C
ENST00000618469.2:c.56A>C
ENST00000634433.2:c.56A>C
ENST00000644379.2:c.56A>C
ENST00000644555.2:c.-231A>C
ENST00000652672.2:c.-205A>C
ENST00000484087.6:c.56A>C
ENST00000700182.1:c.56A>C
ENST00000700183.1:c.56A>C
ENST00000700184.1:n.299A>C
ENST00000700185.1:n.175A>C
ENST00000700186.1:n.175A>C
ENST00000357654.9:c.56A>C
ENST00000471181.7:c.56A>C
ENST00000642945.1:c.56A>C
ENST00000644555.1:c.-231A>C
ENST00000652672.1:c.-205A>C
ENST00000352993.7:c.56A>C
ENST00000354071.7:c.56A>C

ENST00000357654.7:c.56A>C
ENST00000461221.5:c.56A>C
ENST00000461798.5:c.56A>C
ENST00000468300.5:c.56A>C
ENST00000470026.5:c.56A>C
ENST00000471181.6:c.56A>C
ENST00000476777.5:c.56A>C
ENST00000477152.5:c.56A>C
ENST00000478531.5:c.56A>C
ENST00000489037.1:c.56A>C
ENST00000491747.6:c.56A>C
ENST00000492859.5:c.56A>C
ENST00000493795.5:c.-32A>C
ENST00000493919.5:c.-32A>C
ENST00000494123.5:c.56A>C
ENST00000497488.1:c.-219+1230A>C
ENST00000586385.5:c.4+1141A>C
ENST00000591534.5:c.-44+1230A>C
ENST00000591849.5:c.-99+1230A>C
ENST00000618469.1:c.56A>C
ENST00000634433.1:c.56A>C
NM_007294.3:c.56A>C
NM_007297.3:c.-32A>C
NM_007298.3:c.56A>C
NM_007299.3:c.56A>C
NM_007300.3:c.56A>C
NR_027676.1:n.217A>C
NM_007297.4:c.-32A>C
NM_007299.4:c.56A>C
NM_007300.4:c.56A>C
NR_027676.2:n.258A>C

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PS3 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 BRCA1 Version 1.2*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The c.56A>C variant in BRCA1 is a missense variant predicted to cause substitution of Glutamine by Proline at amino acid 19 (p.(Q19P)). This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met). This BRCA1 missense variant is within a key functional domain and a SpliceAI score of 0.03 predicts no impact on splicing (score threshold ≤0.1). The computational predictor

BayesDel (noAF) gives a score of 0.33, above the recommended threshold of 0.28 for prediction of impact on BRCA1 function via protein change (PP3 met). Reported by two calibrated studies to affect protein function similar to pathogenic control variants (PMIDs:30209399, 35659930) (PS3 met). 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 (v1.2) (PS3, PM2_Supporting, PP3).

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met).
PS3	✔	Reported by two calibrated studies to affect protein function similar to pathogenic control variants (PMIDs:30209399, 35659930) (PS3 met).
PP3	✔	This BRCA1 missense variant is within a key functional domain and a SpliceAI score of 0.03 predicts no impact on splicing (score threshold ≤0.1). The computational predictor BayesDel (noAF) gives a score of 0.33, above the recommended threshold of 0.28 for prediction of impact on BRCA1 function via protein change (PP3 met).

Curation History

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.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.