

Variant: NM_007294.4(BRCA1):c.4987-7A>G

Version: 1.0

CA916080172 [↗](#)

868490 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: ac56d2a6-5652-42cf-8d1a-6aaebaca1be8

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HGVS expressions

NM_007294.4:c.4987-7A>G

- NM_007294.4(BRCA1):c.4987-7A>G
- NC_000017.11:g.43067702T>C
- CM000679.2:g.43067702T>C
- NC_000017.10:g.41219719T>C
- CM000679.1:g.41219719T>C
- NC_000017.9:g.38473245T>C
- NG_005905.2:g.150282A>G
- ENST00000461574.2:c.4984-7A>G
- ENST00000470026.6:c.4987-7A>G
- ENST00000473961.6:c.4861-7A>G
- ENST00000476777.6:c.4981-7A>G
- ENST00000477152.6:c.4909-7A>G
- ENST00000478531.6:c.1675-7A>G
- ENST00000489037.2:c.4909-7A>G
- ENST00000493919.6:c.1537-7A>G
- ENST00000494123.6:c.4987-7A>G
- ENST00000497488.2:c.4099-7A>G
- ENST00000618469.2:c.4987-7A>G
- ENST00000634433.2:c.4864-7A>G
- ENST00000644379.2:c.5053-7A>G
- ENST00000644555.2:c.1537-7A>G
- ENST00000652672.2:c.4846-7A>G
- ENST00000484087.6:c.1549-7A>G
- ENST00000357654.9:c.4987-7A>G
- ENST00000471181.7:c.5050-7A>G
- ENST00000644379.1:c.1374-7A>G
- ENST00000352993.7:c.1561-7A>G
- ENST00000357654.7:c.4987-7A>G
- ENST00000461221.5:c.*4770-7A>G
- ENST00000468300.5:c.1675-7A>G
- ENST00000471181.6:c.5050-7A>G
- ENST00000472490.1:n.140-7A>G
- ENST00000478531.5:c.1675-7A>G
- ENST00000484087.5:c.1300-7A>G
- ENST00000491747.6:c.1675-7A>G
- ENST00000493795.5:c.4846-7A>G

ENST00000493919.5:c.1537-7A>G
ENST00000586385.5:c.5-3751A>G
ENST00000591534.5:c.460-7A>G
ENST00000591849.5:c.-98-17512A>G
NM_007294.3:c.4987-7A>G
NM_007297.3:c.4846-7A>G
NM_007298.3:c.1675-7A>G
NM_007299.3:c.1675-7A>G
NM_007300.3:c.5050-7A>G
NR_027676.1:n.5123-7A>G
NM_007297.4:c.4846-7A>G
NM_007299.4:c.1675-7A>G
NM_007300.4:c.5050-7A>G
NR_027676.2:n.5164-7A>G

Likely Pathogenic

Met criteria codes **3**

PM2 PS3 PP3

Not Met criteria codes **1**

PS1

Evidence Links **1**

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 1.2.0*

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

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

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The c.4987-7A>G variant is an intronic variant occurring in intron 15 of the BRCA1 gene. 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 intronic variant is located outside of the native donor and acceptor 1,2 splice sites, and has a SpliceAI score of 0.65, predicting an impact on splicing (score threshold ≥ 0.2) (PP3 met). This intronic variant has functional data from an assay that measures the effect via mRNA and protein. It was reported by one calibrated study incorporating mRNA splicing effects to exhibit protein function similar to pathogenic control variants (PMID:30209399) (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 (PM2_supporting, PP3, PS3).

Met criteria codes

PM2



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).

PS3



Intronic variant, functional data considered only from assays that measure effect via mRNA and protein. Reported by one calibrated study incorporating mRNA splicing effects to exhibit protein function similar to pathogenic control variants (PMID:30209399) (PS3 met).

non functional [PubMed:30209399](#)

PP3



This BRCA1 intronic variant is located outside of the native donor and acceptor 1,2 splice sites, and has a SpliceAI score of 0.65, predicting an impact on splicing (score threshold ≥ 0.2).

Not Met criteria codes

PS1



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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