

Variant: *NM_007294.4(BRCA1):c.70T>A (p.Cys24Ser)*

Version: 1.0

CA10602017 [↗](#)

638952 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d31081e6-bce5-4959-a064-c4e916b0115d

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HGVS expressions

NM_007294.4:c.70T>A

NM_007294.4(BRCA1):c.70T>A (p.Cys24Ser)

NC_000017.11:g.43124027A>T

CM000679.2:g.43124027A>T

NC_000017.10:g.41276044A>T

CM000679.1:g.41276044A>T

NC_000017.9:g.38529570A>T

NG_005905.2:g.93957T>A

ENST00000354071.8:n.134T>A

ENST00000461574.2:c.70T>A

ENST00000470026.6:c.70T>A

ENST00000473961.6:c.70T>A

ENST00000476777.6:c.70T>A

ENST00000477152.6:c.70T>A

ENST00000478531.6:c.70T>A

ENST00000489037.2:c.70T>A

ENST00000493919.6:c.-18T>A

ENST00000494123.6:c.70T>A

ENST00000497488.2:c.-219+1244T>A

ENST00000618469.2:c.70T>A

ENST00000634433.2:c.70T>A

ENST00000644379.2:c.70T>A

ENST00000644555.2:c.-217T>A

ENST00000652672.2:c.-191T>A

ENST00000484087.6:c.70T>A

ENST00000700182.1:c.70T>A

ENST00000700183.1:c.70T>A

ENST00000700184.1:n.313T>A

ENST00000700185.1:n.189T>A

ENST00000700186.1:n.189T>A

ENST00000357654.9:c.70T>A

ENST00000471181.7:c.70T>A

ENST00000642945.1:c.70T>A

ENST00000644555.1:c.-217T>A

ENST00000652672.1:c.-191T>A

ENST00000352993.7:c.70T>A

ENST00000354071.7:c.70T>A

ENST00000357654.7:c.70T>A
ENST00000461221.5:c.70T>A
ENST00000461798.5:c.70T>A
ENST00000468300.5:c.70T>A
ENST00000470026.5:c.70T>A
ENST00000471181.6:c.70T>A
ENST00000476777.5:c.70T>A
ENST00000477152.5:c.70T>A
ENST00000478531.5:c.70T>A
ENST00000489037.1:c.70T>A
ENST00000491747.6:c.70T>A
ENST00000492859.5:c.70T>A
ENST00000493795.5:c.-18T>A
ENST00000493919.5:c.-18T>A
ENST00000494123.5:c.70T>A
ENST00000497488.1:c.-219+1244T>A
ENST00000586385.5:c.4+1155T>A
ENST00000591534.5:c.-44+1244T>A
ENST00000591849.5:c.-99+1244T>A
ENST00000618469.1:c.70T>A
ENST00000634433.1:c.70T>A
NM_007294.3:c.70T>A
NM_007297.3:c.-18T>A
NM_007298.3:c.70T>A
NM_007299.3:c.70T>A
NM_007300.3:c.70T>A
NR_027676.1:n.231T>A
NM_007297.4:c.-18T>A
NM_007299.4:c.70T>A
NM_007300.4:c.70T>A
NR_027676.2:n.272T>A

Likely Pathogenic

Met criteria codes 3

PS3 PP3 PM2_Supporting

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 BRCA2 Version 1.1.0
- Criteria Specification Approval History
- Criteria Specifications for this VCEP

Evidence submitted by expert panel

ENIGMA BRCA1 and BRCA2 VCEP

The variant c.70T>A in BRCA1 is a missense variant predicted to cause substitution of Cysteine by Serine at amino acid 24 (p.Cys24Ser). 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 the computational predictor BayesDel (noAF) gives a score of 0.56, above the recommended threshold of 0.28

for prediction of impact on BRCA1 function via protein change. A SpliceAI score of 0.02 predicts no impact on splicing (score threshold <0.10) (PP3 met). This variant is reported by one calibrated study 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 (PS3, PM2_Supporting, PP3).

Met criteria codes

PS3			Reported by one calibrated study to affect protein function similar to pathogenic control variants (PMID:30209399).
Variant classified as non-functional based on a saturation genome editing (SGE) assay measuring survival in a haploid cell line. PubMed:30209399 			
PP3			This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.56, above the recommended threshold of 0.28 for prediction of impact on BRCA1 function via protein change. A SpliceAI score of 0.02 predicts no impact on splicing (score threshold <0.10) (PP3 met).
PM2_Supporting			This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met).

Curation History



 

 

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