

Variant: NM_007294.4(BRCA1):c.71G>A (p.Cys24Tyr)

Version: 1.0

CA003834 [↗](#)

55678 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 95473e55-2cca-4f71-b173-487bf2dda037

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HGVS expressions

NM_007294.4:c.71G>A

NM_007294.4(BRCA1):c.71G>A (p.Cys24Tyr)

NC_000017.11:g.43124026C>T

CM000679.2:g.43124026C>T

NC_000017.10:g.41276043C>T

CM000679.1:g.41276043C>T

NC_000017.9:g.38529569C>T

NG_005905.2:g.93958G>A

ENST00000354071.8:n.135G>A

ENST00000461574.2:c.71G>A

ENST00000470026.6:c.71G>A

ENST00000473961.6:c.71G>A

ENST00000476777.6:c.71G>A

ENST00000477152.6:c.71G>A

ENST00000478531.6:c.71G>A

ENST00000489037.2:c.71G>A

ENST00000493919.6:c.-17G>A

ENST00000494123.6:c.71G>A

ENST00000497488.2:c.-219+1245G>A

ENST00000618469.2:c.71G>A

ENST00000634433.2:c.71G>A

ENST00000644379.2:c.71G>A

ENST00000644555.2:c.-216G>A

ENST00000652672.2:c.-190G>A

ENST00000484087.6:c.71G>A

ENST00000700182.1:c.71G>A

ENST00000700183.1:c.71G>A

ENST00000700184.1:n.314G>A

ENST00000700185.1:n.190G>A

ENST00000700186.1:n.190G>A

ENST00000357654.9:c.71G>A

ENST00000471181.7:c.71G>A

ENST00000642945.1:c.71G>A

ENST00000644555.1:c.-216G>A

ENST00000652672.1:c.-190G>A

ENST00000352993.7:c.71G>A

ENST00000354071.7:c.71G>A

ENST00000357654.7:c.71G>A
ENST00000461221.5:c.71G>A
ENST00000461798.5:c.71G>A
ENST00000468300.5:c.71G>A
ENST00000470026.5:c.71G>A
ENST00000471181.6:c.71G>A
ENST00000476777.5:c.71G>A
ENST00000477152.5:c.71G>A
ENST00000478531.5:c.71G>A
ENST00000489037.1:c.71G>A
ENST00000491747.6:c.71G>A
ENST00000492859.5:c.71G>A
ENST00000493795.5:c.-17G>A
ENST00000493919.5:c.-17G>A
ENST00000494123.5:c.71G>A
ENST00000497488.1:c.-219+1245G>A
ENST00000586385.5:c.4+1156G>A
ENST00000591534.5:c.-44+1245G>A
ENST00000591849.5:c.-99+1245G>A
ENST00000618469.1:c.71G>A
ENST00000634433.1:c.71G>A
NM_007294.3:c.71G>A
NM_007297.3:c.-17G>A
NM_007298.3:c.71G>A
NM_007299.3:c.71G>A
NM_007300.3:c.71G>A
NR_027676.1:n.232G>A
NM_007297.4:c.-17G>A
NM_007299.4:c.71G>A
NM_007300.4:c.71G>A
NR_027676.2:n.273G>A

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PS3 PP3

Not Met criteria codes 3

BP5 PS1 PP4

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.71G>A variant in BRCA1 is a missense variant predicted to cause substitution of Cys by Tyr at amino acid 24 (p.Cys24Tyr). This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.55, 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 ≤0.1) (PP3 met). This variant is absent from gnomAD v4.1 (read depth ≥25x in >90% samples, PM2_Supporting met). Reported by two calibrated studies to affect protein function similar to pathogenic control variants

(PMIDs:30209399, 35659930) (PS3 met). Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 1.33 (based on Family History LR=1.33), which is above the ENIGMA BRCA1/2 VCEP threshold for BP5 (>0.48) and below PP4 (<2.08) (BP5 and PP4 not 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 (PS3, PP3, PM2_Supporting).

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 the computational predictor BayesDel (noAF) gives a score of 0.55, 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 ≤0.1) (PP3 met).

Not Met criteria codes

BP5			Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 1.33 (based on Family History LR=1.33), which is above the ENIGMA BRCA1/2 VCEP threshold for BP5 (>0.48) and below PP4 (<2.08) (BP5 and PP4 not met; PMID: 31853058).
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			Multifactorial likelihood ratio analysis using clinically calibrated data produced a combined LR for this variant of 1.33 (based on Family History LR=1.33), which is above the ENIGMA BRCA1/2 VCEP threshold for BP5 (>0.48) and below PP4 (<2.08) (BP5 and PP4 not met; PMID: 31853058).

Curation History

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