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Variant: NM_007294.4(BRCA1):c.5138T>C (p.Val1713Ala)

Version: 1.0

CA003262

55412 (ClinVar)

Gene: BRCA1 (HGNC:672)
Condition: BRCA1-related cancer predisposition (MONDO:0700268)
Inheritance Mode: Autosomal dominant inheritance
UUID: b1620e3c-83ab-4f70-9d1d-b2d14d5c7785
Approved on: 2025/12/3
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HGVS expressions

NM_007294.4(BRCA1):c.5138T>C (p.Val1713Ala)
NM_007294.4:c.5138T>C
NC_000017.11:g.43063888A>G
CM000679.2:g.43063888A>G
NC_000017.10:g.41215905A>G
CM000679.1:g.41215905A>G
NC_000017.9:g.38469431A>G
NG_005905.2:g.154096T>C
ENST00000461574.2:c.5135T>C
ENST00000470026.6:c.5138T>C
ENST00000473961.6:c.5012T>C
ENST00000476777.6:c.5132T>C
ENST00000477152.6:c.5060T>C
ENST00000478531.6:c.1826T>C
ENST00000489037.2:c.5060T>C
ENST00000493919.6:c.1688T>C
ENST00000494123.6:c.5138T>C
ENST00000497488.2:c.4250T>C
ENST00000618469.2:c.5138T>C
ENST00000634433.2:c.5015T>C
ENST00000644379.2:c.5204T>C
ENST00000644555.2:c.1688T>C
ENST00000652672.2:c.4997T>C
ENST00000484087.6:c.1700T>C
ENST00000357654.9:c.5138T>C
ENST00000471181.7:c.5201T>C
ENST00000644379.1:c.1525T>C
ENST00000352993.7:c.1712T>C
ENST00000357654.7:c.5138T>C
ENST00000461221.5:c.*4921T>C
ENST00000468300.5:c.1826T>C
ENST00000471181.6:c.5201T>C
ENST00000478531.5:c.1826T>C
ENST00000484087.5:c.1451T>C
ENST00000491747.6:c.1826T>C
ENST00000493795.5:c.4997T>C
ENST00000493919.5:c.1688T>C

ENST00000586385.5:c.68T>C
ENST00000591534.5:c.611T>C
ENST00000591849.5:c.-98-13698T>C
NM_007294.3:c.5138T>C
NM_007297.3:c.4997T>C
NM_007298.3:c.1826T>C
NM_007299.3:c.1826T>C
NM_007300.3:c.5201T>C
NR_027676.1:n.5274T>C
NM_007297.4:c.4997T>C
NM_007299.4:c.1826T>C
NM_007300.4:c.5201T>C
NR_027676.2:n.5315T>C

Likely Pathogenic

Met criteria codes 3

PS3 PM2_Supporting 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.5138T>C variant in BRCA1 is a missense variant predicted to cause substitution of Valine by Alanine at amino acid 1713 (p.(Val1713Ala)). This variant is absent from gnomAD v4.1 (read depth ≥25) (PM2_Supporting met). Reported by four calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:30209399, 30257991, 38709234, 35196514) (PS3 met). This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.4, above the recommended threshold of 0.28 for prediction of impact on BRCA1 function via protein change. A SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score threshold 0.10-0.20) (PP3 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, PS3, PP3).

Met criteria codes

PS3	✔	Reported by four calibrated studies to exhibit protein function similar to pathogenic control variants (PMIDs:30209399, 30257991, 38709234, 35196514) (PS3 met).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1 (read depth ≥25) (PM2_Supporting met).
PP3	✔	This BRCA1 missense variant is within a key functional domain and the computational predictor BayesDel (noAF) gives a score of 0.4, above the recommended threshold of 0.28 for prediction of impact on BRCA1 function via protein change. A SpliceAI score of 0.14 indicates an unclear predicted impact on splicing (score threshold 0.10-0.20) (PP3 met).

Curation History

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