

Variant: *NM_000059.4:c.5632_5634del*

Version: 2.0

CA022758 [↗](#)

193790 (ClinVar) [↗](#)

Gene: BRCA2 ([HGNC:675](#))

Condition: BRCA2-related cancer predisposition ([MONDO:0012933](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: db821c01-c24e-4cd0-a4ab-97da70f8fb35

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HGVS expressions

NM_000059.4:c.5632_5634del

NC_000013.11:g.32339987_32339989del

CM000675.2:g.32339987_32339989del

NC_000013.10:g.32914124_32914126del

CM000675.1:g.32914124_32914126del

NC_000013.9:g.31812124_31812126del

NG_012772.3:g.29508_29510del

ENST00000470094.2:c.5632_5634del

ENST00000528762.2:c.5632_5634del

ENST00000530893.7:c.5263_5265del

ENST00000665585.2:c.5632_5634del

ENST00000666593.2:c.5632_5634del

ENST00000700202.2:c.5632_5634del

ENST00000380152.8:c.5632_5634del

ENST00000544455.6:c.5632_5634del

ENST00000614259.2:c.5632_5634del

ENST00000680887.1:c.5632_5634del

ENST00000380152.7:c.5632_5634del

ENST00000544455.5:c.5632_5634del

ENST00000614259.1:n.5632_5634del

NM_000059.3:c.5632_5634del

Likely Benign

Met criteria codes **1**

BP1_Strong

Not Met criteria codes **3**

PM2

BS1

BA1

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 BRCA2 Version 1.0.0

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

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

ENIGMA BRCA1 and BRCA2 VCEP

The c.5632_5634del variant in BRCA2 is predicted to cause a change in the length of the protein due to an in-frame deletion of one amino acid (p.Asn1878del). This variant is present in gnomAD v2.1 (exomes only, non-cancer subset) or gnomAD v3.1 (non-cancer subset) but is below the ENIGMA BRCA1/2 VCEP threshold >0.00002 for BS1_Supporting (PM2_Supporting, BS1, and BA1 are not met). This in-frame deletion variant is located outside of a key functional domain and was not predicted to alter mRNA splicing using the SpliceAI predictor (score 0.01, score threshold <0.1) (BP1_Strong met). In summary, this variant meets the criteria to be classified as a Likely benign variant for BRCA2-related cancer predisposition based on the ACMG/AMP criteria applied as specified by the ENIGMA BRCA1/2 VCEP (BP1_Strong).

Met criteria codes

BP1_Strong			This in-frame deletion variant is located outside of a key functional domain and was not predicted to alter mRNA splicing using the SpliceAI predictor (score 0.01, score threshold <0.1) (BP1_Strong met).
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Not Met criteria codes

PM2			This variant is present in gnomAD v2.1 (exomes only, non-cancer subset) or gnomAD v3.1 (non-cancer subset) but is below the ENIGMA BRCA1/2 VCEP threshold >0.00002 for BS1_Supporting (PM2_Supporting, BS1, and BA1 are not met).
BS1			This variant is present in gnomAD v2.1 (exomes only, non-cancer subset) or gnomAD v3.1 (non-cancer subset) but is below the ENIGMA BRCA1/2 VCEP threshold >0.00002 for BS1_Supporting (PM2_Supporting, BS1, and BA1 are not met).
BA1			This variant is present in gnomAD v2.1 (exomes only, non-cancer subset) or gnomAD v3.1 (non-cancer subset) but is below the ENIGMA BRCA1/2 VCEP threshold >0.00002 for BS1_Supporting (PM2_Supporting, BS1, and BA1 are not met).

Curation History



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