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Variant: NM_007294.4(BRCA1):c.154C>T (p.Leu52Phe)

Version: 1.0

CA001039

37421 (ClinVar)

Gene: BRCA1 (HGNC:672)
Condition: BRCA1-related cancer predisposition (MONDO:0700268)
Inheritance Mode: Autosomal dominant inheritance
UUID: b6e085c9-0306-45c6-bca2-100ed3cf54e6
Approved on: 2026/7/15
Published on: 2026/7/15

HGVS expressions

NM_007294.4(BRCA1):c.154C>T (p.Leu52Phe)
NM_007294.4:c.154C>T
NC_000017.11:g.43106514G>A
CM000679.2:g.43106514G>A
NC_000017.10:g.41258531G>A
CM000679.1:g.41258531G>A
NC_000017.9:g.38512057G>A
NG_005905.2:g.111470C>T
ENST00000354071.8:n.218C>T
ENST00000461574.2:c.154C>T
ENST00000470026.6:c.154C>T
ENST00000473961.6:c.154C>T
ENST00000476777.6:c.154C>T
ENST00000477152.6:c.135-1558C>T
ENST00000478531.6:c.154C>T
ENST00000489037.2:c.135-1558C>T
ENST00000493919.6:c.13C>T
ENST00000494123.6:c.154C>T
ENST00000497488.2:c.-218-11654C>T
ENST00000618469.2:c.154C>T
ENST00000634433.2:c.154C>T
ENST00000644379.2:c.154C>T
ENST00000644555.2:c.13C>T
ENST00000652672.2:c.13C>T
ENST00000484087.6:c.154C>T
ENST00000700182.1:c.135-1558C>T
ENST00000700183.1:c.*90C>T
ENST00000700184.1:n.397C>T
ENST00000357654.9:c.154C>T
ENST00000471181.7:c.154C>T
ENST00000642945.1:c.*28C>T
ENST00000644555.1:c.13C>T
ENST00000652672.1:c.13C>T
ENST00000352993.7:c.154C>T
ENST00000354071.7:c.154C>T
ENST00000357654.7:c.154C>T
ENST00000461221.5:c.154C>T

ENST00000461798.5:c.154C>T
ENST00000468300.5:c.154C>T
ENST00000470026.5:c.154C>T
ENST00000471181.6:c.154C>T
ENST00000476777.5:c.154C>T
ENST00000477152.5:c.135-1558C>T
ENST00000478531.5:c.154C>T
ENST00000489037.1:c.135-1558C>T
ENST00000491747.6:c.154C>T
ENST00000492859.5:c.*90C>T
ENST00000493795.5:c.13C>T
ENST00000493919.5:c.13C>T
ENST00000494123.5:c.154C>T
ENST00000497488.1:c.-218-11654C>T
ENST00000586385.5:c.4+18668C>T
ENST00000591534.5:c.-44+18757C>T
ENST00000591849.5:c.-99+18757C>T
ENST00000634433.1:c.154C>T
NM_007294.3:c.154C>T
NM_007297.3:c.13C>T
NM_007298.3:c.154C>T
NM_007299.3:c.154C>T
NM_007300.3:c.154C>T
NR_027676.1:n.315C>T
NM_007297.4:c.13C>T
NM_007299.4:c.154C>T
NM_007300.4:c.154C>T
NR_027676.2:n.356C>T

Benign

Met criteria codes 1

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 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.154C>T variant in BRCA1 is a missense variant predicted to cause substitution of Leucine by Phenylalanine at amino acid 52 (p.(Leu52Phe)). The Total GrpMax filtering allele frequency (the lower threshold of the 95% CI) in gnomAD v4.1 is 0.001942 in the East Asian genetic ancestry group (based on 103/44742 alleles) which is above the ENIGMA BRCA1/2 VCEP threshold (>0.001) for BA1 (BA1 met). In

summary, this variant meets the criteria to be classified as a Benign variant for BRCA1-related cancer predisposition based on the ACMG/AMP criteria applied as specified by the ENIGMA BRCA1/2 VCEP (v1.2) (BA1).

Met criteria codes

BA1	✔	The Total GrpMax filtering allele frequency (the lower threshold of the 95% CI) in gnomAD v4.1 is 0.001942 in the East Asian genetic ancestry group (based on 103/44742 alleles) which is above the ENIGMA BRCA1/2 VCEP threshold (>0.001) for BA1 (BA1 met).

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_007294.4(BRCA1):c.154C>T (p.Leu...	Benign	BRCA1-Related Cancer Predisposition	2026-07-15	1.0	ClinGen ENIGMA BRCA1 and BRCA2 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for BRCA1 Version 1.2.0	BRCA1

Showing 1 to 1 of 1 rows

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