

Product datasheet for **RC402818**

BRCA1 (NM_007294) Human Mutant ORF Clone

Product data:

Product Type:	Mutant ORF Clones
Product Name:	BRCA1 (NM_007294) Human Mutant ORF Clone
Mutation Description:	Q19X
Affected Codon#:	19
Affected NT#:	55
Nucleotide Mutation:	BRCA1 Mutant (Q19X), Myc-DDK-tagged ORF clone of Homo sapiens breast Cancer, early onset (BRCA1), transcript variant 1 as transfection-ready DNA
Effect:	Breast and/or ovarian cancer
Symbol:	BRCA1
Synonyms:	BRCAI; BRCC1; BROVCA1; FANCS; IRIS; PNCA4; PPP1R53; PSCP; RNF53
E. coli Selection:	Kanamycin (25 ug/mL)
Mammalian Cell Selection:	Neomycin
Vector:	pCMV6-Entry (PS100001)
Tag:	Myc-DDK
ACCN:	NM_007294
ORF Size:	54 bp
Restriction Sites:	SgfI-MluI
ORF Nucleotide Sequence:	>RC402818 representing NM_007294 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC
GCC**CGATCGCC**

ATGGATTATCTGCTCTTCGCGTTGAAGAAGTACAAAATGTCATTAATGCTATG

AG**CGGACCG**ACGCGTACGCGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
TGGATTACAAGGATGACGACGA TAAGTTTAA



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Locus ID: 672
Cytogenetics: 17q21.31
Domains: BRCT, RING

Protein Families: Druggable Genome, Transcription Factors

Protein Pathways: Ubiquitin mediated proteolysis

MW: 2 kDa

Gene Summary: This gene encodes a 190 kD nuclear phosphoprotein that plays a role in maintaining genomic stability, and it also acts as a tumor suppressor. The BRCA1 gene contains 22 exons spanning about 110 kb of DNA. The encoded protein combines with other tumor suppressors, DNA damage sensors, and signal transducers to form a large multi-subunit protein complex known as the BRCA1-associated genome surveillance complex (BASC). This gene product associates with RNA polymerase II, and through the C-terminal domain, also interacts with histone deacetylase complexes. This protein thus plays a role in transcription, DNA repair of double-stranded breaks, and recombination. Mutations in this gene are responsible for approximately 40% of inherited breast cancers and more than 80% of inherited breast and ovarian cancers. Alternative splicing plays a role in modulating the subcellular localization and physiological function of this gene. Many alternatively spliced transcript variants, some of which are disease-associated mutations, have been described for this gene, but the full-length nature of only some of these variants has been described. A related pseudogene, which is also located on chromosome 17, has been identified. [provided by RefSeq, May 2020]