

## Product datasheet for **RC403048**

### 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:	E1250X
Affected Codon#:	1250
Affected NT#:	3748
Nucleotide Mutation:	BRCA1 Mutant (E1250X), Myc-DDK-tagged ORF clone of Homo sapiens breast Cancer, early onset (BRCA1), transcript variant 1 as transfection-ready DNA
Effect:	Breast cancer
Symbol:	BRCA1
Synonyms:	BRCA1; 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:	3747 bp
Restriction Sites:	SgfI-MluI
ORF Nucleotide Sequence:	>RC403048 representing NM_007294 Red=Cloning site Blue=ORF Green=Tags(s)

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**Protein Sequence:** >RC403048 representing NM\_007294  
 Red=Cloning site Green=Tags(s)

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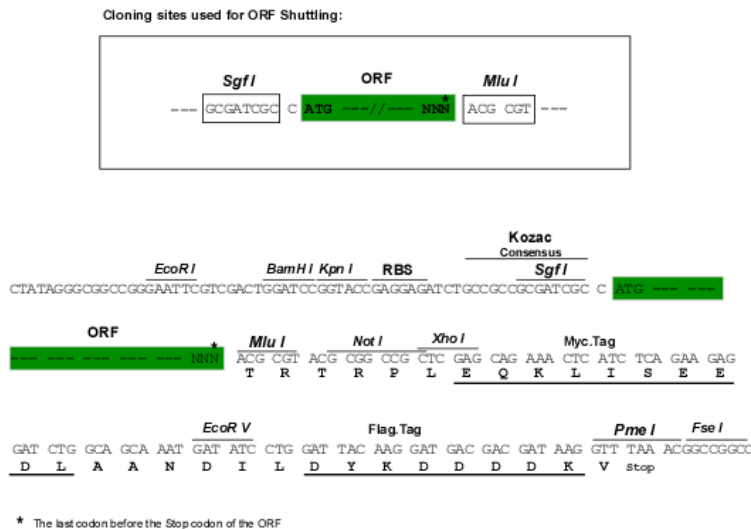
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SGP TRRRLEQKLI SEEDLAANDILDYKDDDDKV

**Restriction Sites:**

SgfI-MluI

**Cloning Scheme:**



**OTI Disclaimer:** Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at [custsupport@origene.com](mailto:custsupport@origene.com) or by calling 301.340.3188 option 3 for pricing and delivery.

The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. [More info](#)

**OTI Annotation:** This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene.

**Components:** The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

**RefSeq:** [NP\\_009225](#)

**RefSeq Size:** 3747 bp

**RefSeq ORF:** 5592 bp

**Locus ID:** 672

**Cytogenetics:** 17q21.31

**Domains:** BRCT, RING

**Protein Families:** Druggable Genome, Transcription Factors

**Protein Pathways:** Ubiquitin mediated proteolysis

**MW:** 137.4 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 natures 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]