

Product datasheet for **RC401540**

MLH1 (NM_000249) Human Mutant ORF Clone

Product data:

Product Type:	Mutant ORF Clones
Product Name:	MLH1 (NM_000249) Human Mutant ORF Clone
Mutation Description:	S193X
Affected Codon#:	193
Affected NT#:	578
Nucleotide Mutation:	MLH1 Mutant (S193X), Myc-DDK-tagged ORF clone of Homo sapiens mutL homolog 1, colon cancer, nonpolyposis type 2 (E. coli) (MLH1), transcript variant 1 as transfection-ready DNA
Effect:	Colorectal cancer, non-polyposis
Symbol:	MLH1
Synonyms:	COCA2; FCC2; hMLH1; HNPCC; HNPCC2; MMRCS1
E. coli Selection:	Kanamycin (25 ug/mL)
Mammalian Cell Selection:	Neomycin
Vector:	pCMV6-Entry (PS100001)
Tag:	Myc-DDK
ACCN:	NM_000249
ORF Size:	576 bp
Restriction Sites:	Sgfl-MluI



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OTI Disclaimer:	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_000240
RefSeq Size:	576 bp
RefSeq ORF:	2271 bp
Locus ID:	4292
Cytogenetics:	3p22.2
Domains:	DNA_mis_repair, HATPase_c
Protein Families:	Druggable Genome
Protein Pathways:	Colorectal cancer, Endometrial cancer, Mismatch repair, Pathways in cancer
MW:	21.1 kDa
Gene Summary:	The protein encoded by this gene can heterodimerize with mismatch repair endonuclease PMS2 to form MutL alpha, part of the DNA mismatch repair system. When MutL alpha is bound by MutS beta and some accessory proteins, the PMS2 subunit of MutL alpha introduces a single-strand break near DNA mismatches, providing an entry point for exonuclease degradation. The encoded protein is also involved in DNA damage signaling and can heterodimerize with DNA mismatch repair protein MLH3 to form MutL gamma, which is involved in meiosis. This gene was identified as a locus frequently mutated in hereditary nonpolyposis colon cancer (HNPCC). [provided by RefSeq, Aug 2017]