

## Product datasheet for **RC401612**

### 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:     | Q510X  |
| Affected Codon#:          | 510  |
| Affected NT#:             | 1528   |
| Nucleotide Mutation:      | MLH1 Mutant (Q510X), 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:                 | 1527 bp  |
| Restriction Sites:        | SgfI-MluI  |



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**ORF Nucleotide Sequence:**

>RC401612 representing NM\_000249  
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC  
 GCCCGCATCGCC

ATGTCGTTCTGTCGAGGGGTTATTCGGCGGCTGGACGAGACAGTGGTGAACCGCATCGCGCGGGGGAAG  
 TTATCCAGCGGCCAGCTAATGCTATCAAAGAGATGATTGAGAAGTGTGTTAGATGCAAAATCCACAAGTAT  
 TCAAGTGATTGTTAAAGAGGGAGGCTGAAGTTGATTCAGATCCAAGACAATGGCACCGGGATCAGGAAA  
 GAAGATCTGGATATTGTATGTGAAAGGTTCACTACTAGTAAACTGCAGTCCTTTGAGGATTTAGCCAGTA  
 TTTCTACCTATGGCTTCGAGGTGAGGCTTTGGCCAGCATAAGCCATGTGGCTCATGTTACTATTACAAC  
 GAAAACAGCTGATGAAAGTGTGCATACAGAGCAAGTACTCAGATGGAAAAGTAAAAGCCCTCCTAAA  
 CCATGTGCTGGCAATCAAGGGACCCAGATCACGGTGGAGGACCTTTTTTACAACATAGCCACGAGGAGAA  
 AAGCTTTAAAAATCAAAGTGAAGAATATGGGAAAATTTGGAAGTTGTTGGCAGGTATTCAGTACACAA  
 TGCAGGCATTAGTTTCTAGTTAAAAACAAGGAGAGACAGTAGCTGATGTTAGGACACTACCCAATGCC  
 TCAACCGTGGACAATATTCGCTCCATCTTTGAAATGCTGTTAGTCGAGAACTGATAGAAATGGATGTG  
 AGGATAAAACCTAGCCTTCAAATGAATGGTTACATATCCAATGCAAACTACTCAGTGAAGAAGTGCAT  
 CTTCTTACTCTTCATCAACCATCGTCTGGTAGAATCAACTTCCTTGAGAAAAGCCATAGAAAACAGTGTAT  
 GCAGCCTATTTGCCAAAAACACACACCATTCTGTACCTCAGTTTAGAAAATCAGTCCCAGAAATGTGG  
 ATGTTAATGTGCACCCCAAAAGCATGAAGTTCACCTCCTGCACGAGGAGAGCATCCTGGAGCGGGTGCA  
 GCAGCACATCGAGAGCAAGCTCCTGGGCTCAAATTCCTCCAGGATGACTTCACCCAGACTTTGCTACCA  
 GGACTTGCTGGCCCTCTGGGGAGATGGTTAAATCCACAACAAGTCTGACCTCGTCTTCTACTTCTGGAA  
 GTAGTGATAAGGTCTATGCCACCAGATGGTTCGTACAGATTCCCGGGAACAGAAGCTTGATGCATTTCT  
 GCAGCCTCTGAGCAAACCCCTGTCCAGTCAGCCCGAGCCATTGTACAGAGGATAAGACAGATATTTCT  
 AGTGGCAGGGCTAGGCAGCAAGATGAGGAGATGCTTGAACCTCCAGCCCTGCTGAAGTGCTGCCAAAA  
 ATCAGAGCTTGGAGGGGGATACAACAAAGGGGACTTCAGAAAATGTGAGAGAAGAGAGGACCTACTCCAG  
 CAACCCAGAAAAGAGACATCGGGAAGATTCTGATGTGGAATGGTGAAGATGATCCCGAAAAGGAAATG  
 ACTGCAGCTTGTACCCCGGAGAAGGATCATTAACCTCACTAGTGTGTTTGTAGTCTC

AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC  
 TGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:**

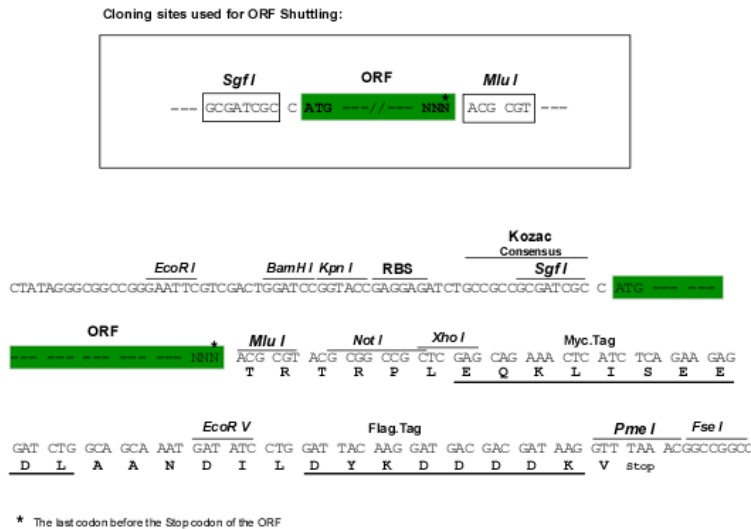
>RC401612 representing NM\_000249  
 Red=Cloning site Green=Tags(s)

MSFVAGVIRRLDETVVNRIAAGEVIQRPANAIEKIENCLDAKSTSIQVIVKEGGLKLIQIQDNGTGIRK  
 EDLDIVCERFTTSKLFQFEDLASISTYGFGEALASISHVAHVITTKTADGKAYRASYSKGKLPKPPK  
 PCAGNQGTQITVEDLFYNIATRRLKKNPSEYKILEVVGRYSVHNAGISFSVKKQGETVADVRLPNA  
 STVDNIRSI FGNAVSRELIEIGCEDKTLAFKMNGYISNANYSVKKCIFLLFINHRLVESTSLRKAIEVY  
 AAYLPKNTHPFLYLSLEISPNQVDVNVHPTKHEVHFLHEESILERVQQHIESKLLGNSSSRMFTQTLPL  
 GLAGPSGEMVKSTTSLTSSSTSGSSDKVYAHQMVRTDSREKLDLQPLSKPLSSQPQAIIVTEKTDIS  
 SGRARQQDEEMLELPAPAEVAANKQSLEGDTTKGTSEMSEKRGPTSSNPKRHRESDVEMVEDDSRKEM  
 TAACTPRRRIINLTSVLSL

SGPTRRRLQKLI SEEDLAANDILDYKDDDDKV

**Restriction Sites:**

Sgfl-MluI

**Cloning Scheme:**

**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:**

1527 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:**

56 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]