

## Product datasheet for **RC401520**

### 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:     | T117R                                                                                                                                                                            |
| Affected Codon#:          | 117                                                                                                                                                                              |
| Affected NT#:             | 350                                                                                                                                                                              |
| Nucleotide Mutation:      | MLH1 Mutant (T117R), 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:                 | 2268 bp                                                                                                                                                                          |
| Restriction Sites:        | SgfI-MluI                                                                                                                                                                        |


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**ORF Nucleotide  
Sequence:**

>RC401520 representing NM\_000249  
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC  
 GCC**CGGATCGCC**

ATGTCGTTCTGTCGAGGGGTTATTTCGGCGGCTGGACGAGACAGTGGTGAACCGCATCGCGCGGGGGAAG  
 TTATCCAGCGGCCAGCTAATGCTATCAAAGAGATGATTGAGAAGTGTGTTAGATGCAAAATCCACAAGTAT  
 TCAAGTGATTGTTAAAGAGGGAGGCCTGAAGTTGATTGATCCAAGACAATGGCACCGGGATCAGGAAA  
 GAAGATCTGGATATTGTATGTGAAAGGTTCACTACTAGTAACTGCAGTCCTTTGAGGATTTAGCCAGTA  
 TTTCTACCTATGGCTTTCGAGGTGAGGCTTTGGCCAGCATAAGCCATGTGGCTCATGTTACTATTACAAG  
 GAAAACAGCTGATGAAAGTGTGCATACAGAGCAAGTTACTCAGATGGAAAAGTAAAGCCCTCCTAAA  
 CCATGTGCTGGCAATCAAGGGACCCAGATCACGGTGGAGGACCTTTTTTACAACATAGCCACGAGGAGAA  
 AAGCTTTAAAAAATCAAGTGAAGAATATGGGAAAAATTTGGAAGTTGTTGGCAGGTATTCAGTACAAA  
 TGCAGGCATTAGTTTCTCAGTAAAAAACAAGGAGAGACAGTAGCTGATGTTAGGACACTACCCAATGCC  
 TCAACCGTGGACAATATTCGCTCCATCTTTGAAATGCTGTTAGTCGAGAACTGATAGAAATGGATGTG  
 AGGATAAAACCTAGCCTTCAAATGAATGGTTACATATCCAATGCAAACTACTCAGTGAAGAAGTGCAT  
 CTTCTTACTCTTCATCAACCATCGTCTGGTAGAATCAACTTCCTTGAGAAAAGCCATAGAAAACAGTGTAT  
 GCAGCCTATTTGCCCAAAACACACACCCATTCTGTACCTCAGTTTAGAAATCAGTCCCCAGAATGTGG  
 ATGTTAATGTGCACCCCAAAAGCATGAAGTTCACCTCCTGCACGAGGAGAGCATCCTGGAGCGGGTGCA  
 GCAGCACATCGAGAGCAAGCTCCTGGGCTCCAATTCTCCAGGATGTACTTCACCCAGACTTTGCTACCA  
 GGACTTGCTGGCCCTCTGGGGAGATGGTTAAATCCACAACAAGTCTGACCTCGTCTTCTACTTCTGGAA  
 GTAGTGATAAGGTCTATGCCCAACAGATGGTTCGTACAGATTCGCGGAACAGAAGCTTGATGCATTTCT  
 GCAGCCTCTGAGCAAACCCCTGTCCAGTCAGCCCCAGGCCATTGTCACAGAGGATAAGACAGATATTTCT  
 AGTGGCAGGGCTAGGCAGCAAGATGAGGAGATGCTTGAACCTCCAGCCCTGCTGAAGTGCTGCCAAAA  
 ATCAGAGCTTGGAGGGGGATACAACAAGGGGACTTCAGAAATGTCAGAGAAGAGAGGACCTACTTCCAG  
 CAACCCAGAAAGAGACATCGGGAAGATTCTGATGTGGAATGGTGAAGATGATTCGGAAGGAAATG  
 ACTGCAGCTTGTACCCCGGAGAAGGATCATTAACCTCACTAGTGTGTTTGAAGTCTCCAGGAAGAAATTA  
 ATGAGCAGGGACATGAGGTTCTCCGGGAGATGTTGCATAACCACTCCTTCGTGGGCTGTGTGAATCCTCA  
 GTGGGCCTTGGCACAGCATCAAACCAAGTTATACCTTCTCAACACCAAGCTTAGTGAAGAACTGTTT  
 TACCAGATACTCATTTATGATTTTGCCAAATTTGGTGTCTCAGGTTATCGGAGCCAGCACCGCTCTTTG  
 ACCTTGCCATGCTTGCTTAGATAGTCCAGAGAGTGGCTGGACAGAGGAAGATGGTCCCAAGAAAGGACT  
 TGCTGAATACATTGTTGAGTTTCTGAAGAAGAAGGCTGAGATGCTTGCACTATTTCTCTTTGGAAAT  
 GATGAGGAAGGGAACCTGATTGGATTACCCCTTCTGATTGACAACTATGTGCCCTTTGGAGGGACTGC  
 CTATCTTCACTTCGACTAGCCACTGAGGTGAATTGGGACGAAGAAAAGGAATGTTTTGAAAGCCTCAG  
 TAAAGAATGCGCTATGTTCTATTCCATCCGGAAGCAGTACATATCTGAGGAGTCGACCCTCTCAGGCCAG  
 CAGAGTGAAGTGCCTGGCTCATTCAAACCTCTGGAAGTGGACTGTGGAACACATTGTCTATAAAGCCT  
 TGCCTCACACATTCTGCCTCTAAACATTTACAGAAGATGGAATATCCTGCAGCTTGCTAACCTGCC  
 TGATCTATACAAAGTCTTGAGAGGTGT

AG**CGGACCG**ACGCGTACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC  
 TGGATTACAAGGATGACGACGA TAAGTTTAA

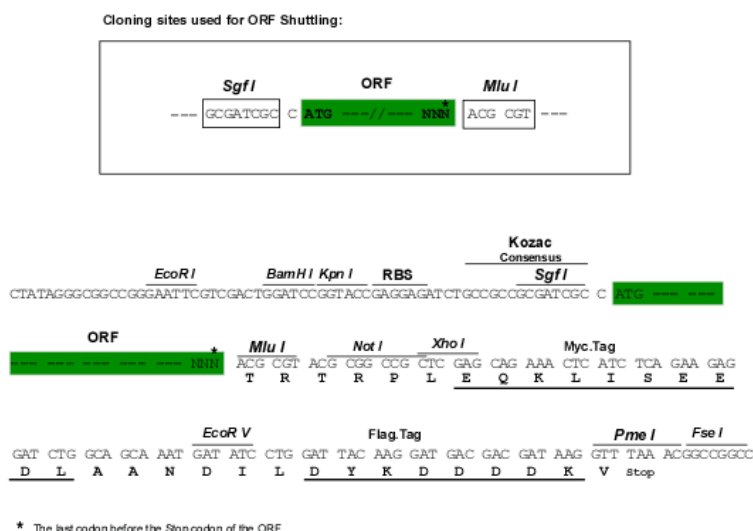
**Protein Sequence:** >RC401520 representing NM\_000249  
Red=Cloning site Green=Tags(s)

MSFVAGVIRRLDETVVNRIAAGEVIQRPANAIAKEMIENCLDAKSTSIQVIVKEGGLKLIQIQDNGTGIRK  
EDLDIVCERFTTSKLQSFEDLASISTYGRGEALASISHVAHVITITRKADGKCAYRASYS DGKLKAPPK  
PCAGNQGTQITVEDLFYNIATRRKALKNPSEEYGKILEVVGGRYSVHNAGISFSVKKQGETVADVRLPNA  
STVDNIRISIFGNAVSRELIEIGCEDKTLAFKMNGYISNANYSVKKCIFLLFINHRLVESTSLRKAJETVY  
AAYLPKNTHPFLYLSLETSPQNVVDVNVHPTKHEVHFLHEESILERVQQHIESKLLGSNSSRMYFTQTLLP  
GLAGPSGEMVKSTTSLTSSSTSGSSDKVYAHQMVRTDSREQKLDAFLQPLSKPLSSQPQAIVTEDKTDIS  
SGRARQQDEEMLELPAPAEVAAKNQSLEGDTTKGTSEMSEKRGPTSSNPRKRHRESDVEMVEDDSRKEM  
TAACTPRRRIINLTSLVLSLQEEINEQGHEVLREMLHNHSFVGCVPNPQWALAQHQTKLYLLNTTKLSEELF  
YQILYIDFANFGLVRLSEPAFLDLAMLALDSPEGWTEEDGPKEGLAEYIVEFLKKAEMLADYFSLEI  
DEEGNLIGLPLLDIDNYVPHLEGLPIFILRLATEVNWDEEKECFESLSEKAMFYSRKKQYISEESTLSGQ  
QSEVPGSPNSKWKTVHEHVVYKALRSHLIPPKHETEDGNTLOLANLPDIYKVFERC

SGPTRRRL EOKLISEEDLAANDILDYKDDDDKV

**Restriction Sites:** SgfI-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.

|                          |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
|--------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Components:</b>       | 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).                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
| <b>Note:</b>             | Plasmids are not sterile. For experiments where strict sterility is required, filtration with 0.22um filter is required.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>RefSeq:</b>           | <u>NP_000240</u>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| <b>RefSeq Size:</b>      | 2268 bp                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>RefSeq ORF:</b>       | 2271 bp                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>Locus ID:</b>         | 4292                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
| <b>Cytogenetics:</b>     | 3p22.2                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>Domains:</b>          | DNA_mis_repair, HATPase_c                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| <b>Protein Families:</b> | Druggable Genome                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| <b>Protein Pathways:</b> | Colorectal cancer, Endometrial cancer, Mismatch repair, Pathways in cancer                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| <b>MW:</b>               | 83.2 kDa                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>Gene Summary:</b>     | 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] |