

## Product datasheet for **RC401683**

### 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:     | W714X                                                                                                                                                                            |
| Affected Codon#:          | 714                                                                                                                                                                              |
| Affected NT#:             | 2142                                                                                                                                                                             |
| Nucleotide Mutation:      | MLH1 Mutant (W714X), 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:                 | 2139 bp                                                                                                                                                                          |
| Restriction Sites:        | SgfI-MluI                                                                                                                                                                        |


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**ORF Nucleotide  
Sequence:**

>RC401683 representing NM\_000249  
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC  
 GCC**CGGATCGCC**

ATGTCGTTCTGTGGCAGGGGTTATTTCGGCGGCTGGACGAGACAGTGGTGAACCGCATCGCGCGGGGGAAG  
 TTATCCAGCGGCCAGCTAATGCTATCAAAGAGATGATTGAGAAGTGTGTTAGATGCAAAATCCACAAGTAT  
 TCAAGTGATTGTTAAAGAGGGAGGCCTGAAGTTGATTGATCCAAGACAATGGCACCGGGATCAGGAAA  
 GAAGATCTGGATATTGTATGTGAAAGGTTCACTACTAGTAACTGCAGTCCTTTGAGGATTTAGCCAGTA  
 TTTCTACCTATGGCTTTCGAGGTGAGGCTTTGGCCAGCATAAGCCATGTGGCTCATGTTACTATTACAAC  
 GAAAACAGCTGATGAAAGTGTGCATACAGAGCAAGTTACTCAGATGGAAAAGTAAAGCCCTCCTAAA  
 CCATGTGCTGGCAATCAAGGGACCCAGATCACGGTGGAGGACCTTTTTTACAACATAGCCACGAGGAGAA  
 AAGCTTTAAAAAATCAAGTGAAGAATATGGGAAAATTTTGAAGTTGTTGGCAGGTATTCAGTACACAA  
 TGCAGGCATTAGTTTCTCAGTTAAAAACAAGGAGAGACAGTAGCTGATGTTAGGACACTACCCAATGCC  
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 CTTCTTACTCTTCATCAACCATCGTCTGGTAGAATCAACTTCCTTGAGAAAAGCCATAGAAAACAGTGTAT  
 GCAGCCTATTTGCCCAAAAACACACACCCATTCTGTACCTCAGTTTAGAAATCAGTCCCCAGAATGTGG  
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 GGACTTGCTGGCCCTCTGGGGAGATGGTTAAATCCACAACAAGTCTGACCTCGTCTTCTACTTCTGGAA  
 GTAGTGATAAGGTCTATGCCCAACAGATGGTTCGTACAGATTCGGGGAACAGAAGCTTGATGCATTTCT  
 GCAGCCTCTGAGCAAACCCCTGTCCAGTCAGCCCCAGGCCATTGTCACAGAGGATAAGACAGATATTTCT  
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 CAACCCAGAAAGAGACATCGGGAAGATTCTGATGTGGAATGGTGAAGATGATTCCCGAAAGGAAATG  
 ACTGCAGCTTGTACCCCGGAGAAGGATCATTAACCTCACTAGTGTGTTTGTAGTCTCCAGGAAGAAATTA  
 ATGAGCAGGGACATGAGGTTCTCCGGGAGATGTTGCATAACCACTCCTTCGTGGGCTGTGTGAATCCTCA  
 GTGGGCTTGGCACAGCATCAAACCAAGTTATACCTTCTCAACACCAAGCTTAGTGAAGAACTGTTT  
 TACCAGATACTCATTTATGATTTTGCCAAATTTGGTGTCTCAGGTTATCGGAGCCAGCACCGCTCTTTG  
 ACCTTGCCATGCTTGCCTTAGATAGTCCAGAGAGTGGCTGGACAGAGGAAGATGGTCCCAAGAAGGACT  
 TGCTGAATACATTGTTGAGTTTCTGAAGAAGAAGGCTGAGATGCTTGCACTATTTCTCTTTGGAAAT  
 GATGAGGAAGGGAACCTGATTGGATTACCCCTTCTGATTGACAACTATGTGCCCTTTGGAGGGACTGC  
 CTATCTTCATTCTCGACTAGCCACTGAGGTGAATTGGGACGAAGAAAAGGAATGTTTTGAAAGCCTCAG  
 TAAAGAATGCGCTATGTTCTATTCCATCCGGAAGCAGTACATATCTGAGGAGTCGACCTCTCAGGCCAG  
 CAGAGTGAAGTGCCTGGCTCCATTCCAACTCCTGGAAG

AG**CGGACCG**ACGCGTACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAATGATATCC  
 TGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:** >RC401683 representing NM\_000249  
 Red=Cloning site Green=Tags(s)

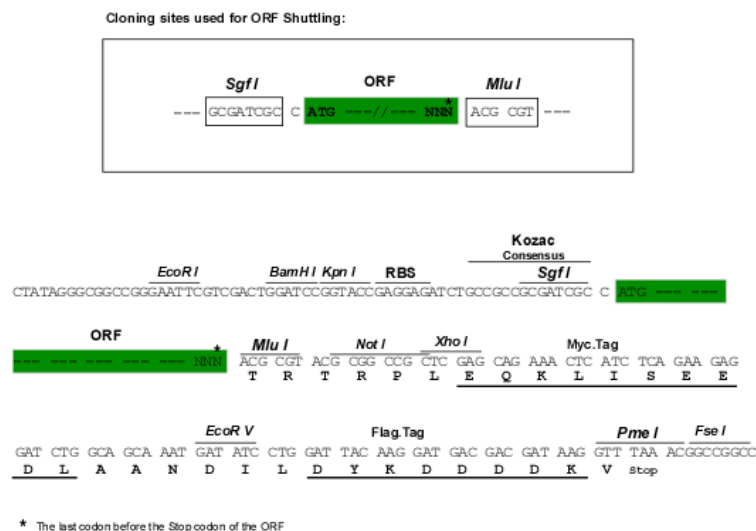
MSFVAGVIRRLDETVVNRIAAGEVIQRPANAIEKMIENCLDAKSTSIQVIVKEGGLKLIQIQDNGTGIRK  
 EDLDIVCERFTTSKLQSFEDLASISTYGRGEALASISHVAHVTTITKTADGKAYRASYSKGKAPPK  
 PCAGNQGTQITVEDLFYNIATRRKALKNPSEEYKILEVVGRYSVHNAGISFSVKKQGETVADVRTLPA  
 STVDNIRSIFGNAVSRELIEIGCEDKTLAFKMNGYISNANYSVKKCIFLLFINHRLVESTSLRKAIVTV  
 AAYLPKNTHPFLYLLEISPNQVNDVNVHPTKHEVHFLHEESILERVQQHIESKLLGSNSSRMYFTQTLLP  
 GLAGPSGEMVKSTTSLTSSSTSGSSDKVYAHQMVRTDSREQKLDALQPLSKPLSSQPQAIVTEKTDIS  
 SGRARQDEEMLELPAPAEVAAKNQSLEGDTTKGTSEMSEKRGPTSSNPRKRHRESDVEMVEDDSRKEM  
 TAACTPRRRIINLTSLVSLQEEINEQGHEVLREMLHNHSFVGCVPQWALAQHQTKLYLLNTTKLSEELF  
 YQILYDFANFGVRLSEAPLFDLAMLALDSPESGWTEEDGPKEGLAEYIVEFLKKKAEMLDYFSLEI  
 DEEGLNIGLPLLDNYVPPLEGLPIFILRLATEVNWDEEKECFESLSKECAMFYISIRKQYISEESTLSGQ  
 QSEVPGSIPNSWK

SGPTRRRLEQKLISEEDLAANDILDYKDDDDKV

**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](#)

|                          |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
|--------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>OTI Annotation:</b>   | 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>      | 2139 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>               | 78.4 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] |