错配修复蛋白 1 抗体

产品货号： mlR23633

英文名称： MLH1

中文名称： 错配修复蛋白 1 抗体

别　　名： mutl homolog l gene; MutL protein homolog 1; DNA mismatch repair protein Mlh1; COCA 2; COCA2; DNA mismatch repair protein Mlh1; FCC 2; FCC2; hMLH 1; hMLH1; HNPCC; HNPCC 2; HNPCC2; MGC5172; MLH 1; MutL homolog 1; MutL homolog 1 colon cancer nonpolyposis type 2; MutL protein homolog 1; MLH1_HUMAN.

研究领域： 肿瘤　免疫学　信号转导　转录调节因子

抗体来源： vRabbit

克隆类型： Polyclonal

交叉反应　： Human, Mouse, Rat, Dog, Pig, Horse, Rabbit,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 85kDa

细胞定位： 细胞核

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human MLH1:131-230/756

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 ° C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20° C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 ° C.

PubMed： PubMed

产品介绍： This gene was identified as a locus frequently mutated in hereditary nonpolyposis colon cancer (HNPCC). It is a human homolog of the E. coli DNA mismatch repair gene mutL, consistent with the characteristic alterations in microsatellite sequences (RER+ phenotype) found in HNPCC. Alternatively spliced transcript variants encoding different isoforms have been described, but their full-length natures have not been determined. SUBUNIT: Heterodimer of MLH1 and PMS2 or MLH1 and MLH3. Part of the BRCA1-associated genome surveillance complex (BASC), which contains BRCA1, MSH2, MSH6, MLH1, ATM, BLM, PMS2 and the RAD50- MRE11-NBS1 protein complex. This association could be a dynamic process changing throughout the cell cycle and within subnuclear domains. Interacts with MBD4. Interacts with EXO1.

Function:

Heterodimerizes with PMS2 to form MutL alpha, a component of the post-replicative DNA mismatch repair system (MMR). DNA repair is initiated by MutS alpha (MSH2-MSH6) or MutS beta (MSH2-MSH6) binding to a dsDNA mismatch, then MutL alpha is recruited to the heteroduplex. Assembly of the MutL-MutS-heteroduplex ternary complex in presence of RFC and PCNA is sufficient to activate endonuclease activity of PMS2. It introduces single-strand breaks near the mismatch and thus generates new entry points for the exonuclease EXO1 to degrade the strand containing the mismatch. DNA methylation would prevent cleavage and therefore assure that only the newly mutated DNA strand is going to be corrected. MutL alpha (MLH1-PMS2) interacts physically with the clamp loader subunits of DNA polymerase III, suggesting that it may play a role to recruit the DNA polymerase III to the site of the MMR. Also implicated in DNA damage signaling, a process which induces cell cycle arrest and can lead to apoptosis in case of major DNA damages. Heterodimerizes with MLH3 to form MutL gamma which plays a role in meiosis.

Subunit:

Heterodimer of MLH1 and PMS2 (MutL alpha), MLH1 and PMS1 (MutL beta) or MLH1 and MLH3 (MutL gamma). Forms a ternary complex with MutS alpha (MSH2-MSH6) or MutS beta (MSH2-MSH3). Part of the BRCA1-associated genome surveillance complex (BASC), which contains BRCA1, MSH2, MSH6, MLH1, ATM, BLM, PMS2 and the RAD50-MRE11-NBS1 protein complex. This association could be a dynamic process changing throughout the cell cycle and within subnuclear domains. Interacts with MBD4. Interacts with EXO1 and MTMR15/FAN1.

Subcellular Location:

Nucleus.

Tissue Specificity:

Colon, lymphocytes, breast, lung, spleen, testis, prostate, thyroid, gall bladder and heart.

DISEASE:

Hereditary non-polyposis colorectal cancer 2 (HNPCC2) [MIM:609310]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. Note=The disease is caused by mutations affecting the gene represented in this entry.

Mismatch repair cancer syndrome (MMRCS) [MIM:276300]: Autosomal dominant disorder characterized by malignant tumors of the brain associated with multiple colorectal adenomas. Skin features include sebaceous cysts, hyperpigmented and cafe au lait spots. Note=The disease is caused by mutations affecting the gene represented in this entry.

Muir-Torre syndrome (MRTES) [MIM:158320]: Rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy. Note=The disease is caused by mutations affecting the gene represented in this entry.

Note=Defects in MLH1 may contribute to lobular carcinoma in situ (LCIS), a non-invasive neoplastic disease of the breast.

Endometrial cancer (ENDMC) [MIM:608089]: A malignancy of endometrium, the mucous lining of the uterus. Most endometrial cancers are adenocarcinomas, cancers that begin in cells that make and release mucus and other fluids. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.

Note=Some epigenetic changes can be transmitted unchanged through the germline (termed 'epigenetic inheritance'). Evidence that this mechanism occurs in humans is provided by the identification of individuals in whom 1 allele of the MLH1 gene is epigenetically silenced throughout the soma (implying a germline event). These individuals are affected by HNPCC but does not have identifiable mutations in MLH1, even though it is silenced, which demonstrates that an epimutation can phenocopy a genetic disease.

Similarity:

Belongs to the DNA mismatch repair mutL/hexB family.

SWISS:

P40692

Gene ID:

4292

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

产品图片