abcam

Product datasheet

Recombinant Human MLH1 protein ab131924

1 Image

Description

Product name	Recombinant Human MLH1 protein
Expression system	Wheat germ
Accession	**P40692**
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MSFVAGVIRRLDETVVNRIAAGEVIQRPANAIKEMIENCLDA KSTSIQVI VKEGGLKLIQIQDNGTGIRKEDLDIVCERFTTSKLQSFEDLA SISTYGFR GEALASISHVAHVTITTKTADGKCAYRASYSDGKLKAPPKP CAGNQGTQI TVEDLFYNIATRRKALKNPSEEYGKILEVVGRYSVHNAGISF SVKKQGET VADVRTLPNASTVDNIRSIFGNAVSRELIEIGCEDKTLAFKM NGYISNAN YSVKKCIFLLFINHRLVESTSLRKAIETVYAAYLPKNTHPFLY LSLEISP QNVDVNVHPTKHEVHFLHEESILERVQQHIESKLLGSNSS RMYFTQTLLP GLAGPSGEMVKSTTSLTSSSTSGSSDKVYAHQMVRTDSR EQKLDAFLQPL SKPLSSQPQAIVTEDKTDISSGRARQQDEEMLELPAPAEV AAKNQSLEGD TTKGTSEMSEKRGPTSSNPRKRHREDSDVEMVEDDSRK EMTAACTPRRRI INLTSVLSLQEEINEQGHEVLREMLHNHSFVGCVNPQWAL AQHQTKLYLL NTTKLSEELFYQILIYDFANFGVLRLSEPAPLFDLAMLALDS PESGWTEE DGPKEGLAEYIVEFLKKKAEMLADYFSLEIDEEGNLIGLPLL IDNYVPPL EGLPIFILRLATEVNWDEEKECFESLSKECAMFYSIRKQYIS EESTLSGQ

QSEVPGSIPNSWKWTVEHIVYKALRSHILPPKHFTEDGNIL
QLANLPDLY KVFERC

Predicted molecular weight	109 kDa including tags
Amino acids	1 to 756
Tags	GST tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab131924** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA SDS-PAGE Western blot
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

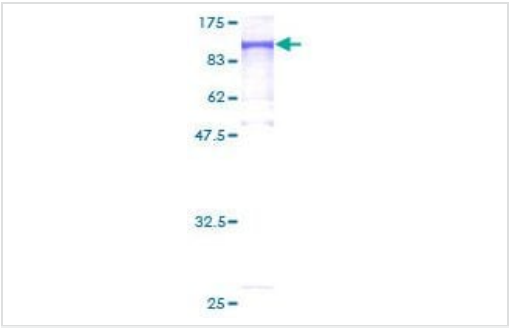
Function	Heterodimerizes with PMS2 to form MutL alpha, a component of the post-replicative DNA mismatch repair system (MMR). DNA repair is initiated by MutS alpha (MSH2-MSH6) or MutS beta (MSH2-MSH6) binding to a dsDNA mismatch, then MutL alpha is recruited to the heteroduplex. Assembly of the MutL-MutS-heteroduplex 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.
Tissue specificity	Colon, lymphocytes, breast, lung, spleen, testis, prostate, thyroid, gall bladder and heart.
Involvement in disease	Defects in MLH1 are the cause of hereditary non-polyposis colorectal cancer type 2 (HNPCC2) [MIM:609310]. Mutations in more than one gene locus can be involved alone or in combination in the production of the HNPCC phenotype (also called Lynch syndrome). Most families with clinically recognized HNPCC have mutations in either MLH1 or MSH2 genes. HNPCC is an autosomal, dominantly inherited disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early onset colorectal carcinoma (CRC) and extra-colonic cancers 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, and accounts for 15% of all colon cancers. Cancers in HNPCC originate within benign

neoplastic polyps termed adenomas. Clinically, HNPCC is often divided into two subgroups. Type I: hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II: patients have an 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.
Defects in MLH1 are a cause of mismatch repair cancer syndrome (MMRCS) [MIM:276300]; also known as Turcot syndrome or brain tumor-polyposis syndrome 1 (BTPS1). MMRCS is an 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.
Defects in MLH1 are a cause of Muir-Torre syndrome (MuToS) [MIM:158320]; also abbreviated MTS. MuToS is a rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy.
Note=Defects in MLH1 may contribute to lobular carcinoma in situ (LCIS), a non-invasive neoplastic disease of the breast.
Defects in MLH1 are a cause of susceptibility to endometrial cancer (ENDMC) [MIM:608089].
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.

Sequence similarities Belongs to the DNA mismatch repair mutL/hexB family.

Cellular localization Nucleus.

Images



SDS-PAGE - Recombinant Human MLH1 protein (ab131924)

12.5% SDS-PAGE analysis of ab131924 stained with Coomassie Blue.

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