

# Recombinant Human MSH2 protein ab114351

画像数 1

### 製品の詳細

製品名	Recombinant Human MSH2 protein
発現系	Wheat germ
アクセッション番号	<b><u>P43246</u></b>
タンパク質長	Full length protein
Animal free	No
由来	Recombinant
生物種	Human
配列	MAVQPKETLQLESAAEVGFVRRFFQGMPEKPTTTVRLFDRGDF YTAHGEDA LLAAREVFKTQGVIKYMGPAKAKNLQSVVLSKMNFEFVKDL LLVRQYRV EYKYNRAGNKASKENDWYLAYKASPGNLSQFEDILFGNNDMS ASIGVVG KMSAVDGGQRQVGVYVDSIQRKLGLCEFPDNDQFSNLEALLI QIGPKECV LPGGETAGDMGKLRQIIQRGGILITERKKADFSTKDIYQDLN RLLKGGKG EQMNSAVLPEMENQVAVSSLSAVIKFLELLSDDSNFGQFELT TFDFSQYM KLDIAAVRALNLFQGSVEDTTGSQSLAALLNKCKTPQGQRLV NQWIKQPL MDKNRIEERLNLVEAFVEDAELRQTLQEDLLRRFPDLNRLAK KFQRQAAN LQDCYRLYQGINQLPNVIQALEKHEGKHQKLLLAVFVTPPLTD LRSDFSKF QEMIETTLDMDQVENHEFLVKPSFDPNLSELREIMNDLEKKM QSTLISAA RDLGLDPGKQIKLDSSAQFGYYFRVTCKEEKVLRNNKNFSTV DIQKNGVK FTNSKLTSLNEEYTKNKTEYEEAQDAIVKEIVNISSGYVEPM QTLNDVLA QLDAVVSFAHVSNGAPVPYVRPAILEKQGRIILKASRHACV EVQDEIAF IPNDVYFEKDKQMFHIIITGPNMGGKSTYIRQTGVIVLMAQIG CFVPCESA

EVSIVDCILARVGAGDSQLKGVSTFMAEMLETASILRSATKD  
SLIIIDEL  
GRGTSTYDGFGLAWAISEYIATKIGAFCMFATHFHELTALAN  
QIPTVNNL  
HVTALTTEETLTMLYQVKKGVCDQSFQIHVAELANFPKHVIE  
CAKQKALE  
LEEFQYIGESQGYDIMEPAAKKCYLEREQGEKIIQEFLSKVK  
QMPFTEMS  
EENITIKLKLKAEVIAKNNSFVNEIISRIKVTT

予測される分子量 129 kDa including tags  
領域 1 to 934

## 特性

Our **Abpromise guarantee** covers the use of **ab114351** in the following tested applications.

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

アプリケーション ELISA  
SDS-PAGE  
Western blot  
製品の状態 Liquid

## 前処理および保存

保存方法および安定性 Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.  
pH: 8.00  
Constituents: 0.3% Glutathione, 0.79% Tris HCl

## 関連情報

機能 Component of the post-replicative DNA mismatch repair system (MMR). Forms two different heterodimers: MutS alpha (MSH2-MSH6 heterodimer) and MutS beta (MSH2-MSH3 heterodimer) which binds to DNA mismatches thereby initiating DNA repair. When bound, heterodimers bend the DNA helix and shields approximately 20 base pairs. MutS alpha recognizes single base mismatches and dinucleotide insertion-deletion loops (IDL) in the DNA. MutS beta recognizes larger insertion-deletion loops up to 13 nucleotides long. After mismatch binding, MutS alpha or beta forms a ternary complex with the MutL alpha heterodimer, which is thought to be responsible for directing the downstream MMR events, including strand discrimination, excision, and resynthesis. ATP binding and hydrolysis play a pivotal role in mismatch repair functions. The ATPase activity associated with MutS alpha regulates binding similar to a molecular switch: mismatched DNA provokes ADP→ATP exchange, resulting in a discernible conformational transition that converts MutS alpha into a sliding clamp capable of hydrolysis-independent diffusion along the DNA backbone. This transition is crucial for mismatch repair. MutS alpha may also play a role in DNA homologous recombination repair. In melanocytes may modulate both UV-B-induced cell cycle regulation and apoptosis.

組織特異性 Ubiquitously expressed.

関連疾患 Defects in MSH2 are the cause of hereditary non-polyposis colorectal cancer type 1 (HNPCC1)

[MIM:120435]. 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. 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. MSH2 mutations may predispose to hematological malignancies and multiple cafe-au-lait spots. Defects in MSH2 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.

Defects in MSH2 are a cause of susceptibility to endometrial cancer (ENDMC) [MIM:608089]. Defects in MSH2 are a cause of hereditary non-polyposis colorectal cancer type 8 (HNPCC8) [MIM:613244]. HNPCC is a 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=HNPCC8 results from heterozygous deletion of 3-prime exons of EPCAM and intergenic regions directly upstream of MSH2, resulting in transcriptional read-through and epigenetic silencing of MSH2 in tissues expressing EPCAM.

#### 配列類似性

Belongs to the DNA mismatch repair mutS family.

#### 翻訳後修飾

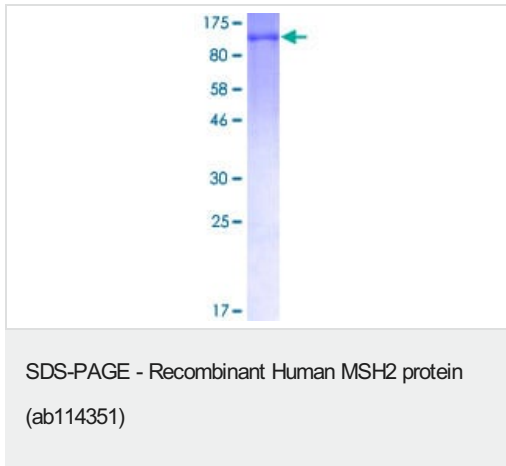
Phosphorylated by PRKCZ, which may prevent MutS alpha degradation by the ubiquitin-proteasome pathway.

Phosphorylated upon DNA damage, probably by ATM or ATR.

#### 細胞内局在

Nucleus.

#### 画像



ab114351 analysed on a 12.5% SDS-PAGE gel stained with Coomassie Blue.

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