abcam

Product datasheet

Recombinant Human MSH6 protein ab152443

1 图像

描述

产品名称 重组人MSH6蛋白

表达系统 Wheat germ

Accession <u>P52701</u>

蛋白长度 Protein fragment

无动物成分 No

性质 Recombinant

种属 Human

序列 AGFDSDYDQALADIRENEQSLLEYLEKQRNRIGCRTIVYWGI

GRNRYQLE

IPENFTTRNLPEEYELKSTKKGCKRYWTKTIEKKLANLINAE

ERRDVSLK

预**测分子量** 37 kDa including tags

氨基酸 931 to 1030

技术指标

Our Abpromise guarantee covers the use of ab152443 in the following tested applications.

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

应**用** ELISA

SDS-PAGE

Western blot

形式 Liquid

补充说明

制备和贮存

稳定性和存储 Shipped on dry ice. Upon delivery aliquot and store at -80℃. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.31% Glutathione, 0.79% Tris HCI

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常规信息

功能

疾病相关

Component of the post-replicative DNA mismatch repair system (MMR). Heterodimerizes with MSH2 to form MutS alpha, which binds to DNA mismatches thereby initiating DNA repair. When bound, MutS alpha bends the DNA helix and shields approximately 20 base pairs, and recognizes single base mismatches and dinucleotide insertion-deletion loops (IDL) in the DNA. After mismatch binding, 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.

Defects in MSH6 are the cause of hereditary non-polyposis colorectal cancer type 5 (HNPCC5) [MIM:600678]. 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. MSH6 mutations appear to be associated with atypical HNPCC and in particular with development of endometrial carcinoma or atypical endometrial hyperplasia, the presumed precursor of endometrial cancer. Defects in MSH6 are also found in familial colorectal cancers (suspected or incomplete HNPCC) that do not fulfill the Amsterdam criteria for HNPCC.

Defects in MSH6 are a cause of susceptibility to endometrial cancer (ENDMC) [MIM:608089].

Belongs to the DNA mismatch repair mutS family.

Contains 1 PWWP domain.

The N-terminus is blocked.

Phosphorylated upon DNA damage, probably by ATM or ATR.

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

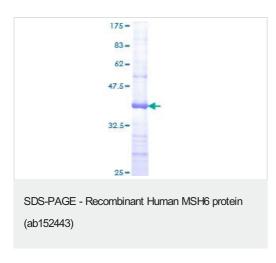
proteasome pathway.

细胞定位 Nucleus.

图片

序列相似性

翻译后修饰



12.5% SDS-PAGE analysis of ab152443 stained with Coomassie Blue.

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