abcam

Product datasheet

Recombinant Human ATM protein ab131739

1 Image

Description

Product name Recombinant Human ATM protein

Purity >= 80 % Purified via GST Tag.

Glutathione Sepharose

Expression system Wheat germ

Protein length Protein fragment

Animal free No

Nature Recombinant

Species Human

Sequence MTLHEPANSSASQSTDLCDFSGDLDPAPNPPHFPSHVV

KATFAYISNCHK

TKLKSILEILSKSPDSYQKILLAICEQAAETNNVYKKHRILKIY

HLFVSL

LLKDIKSGLGGAWAFVLRDVIYTLIHYINQRKLTIFSQ

Predicted molecular weight 41 kDa including tags

Amino acids 1 to 138

Specifications

Our Abpromise guarantee covers the use of ab131739 in the following tested applications.

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

Applications ELISA

Western blot SDS-PAGE

Form Liquid

Additional notes

Preparation and Storage

Stability and Storage Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

Constituents: 0.31% Glutathione, 0.79% Tris HCI

General Info

Function

Serine/threonine protein kinase which activates checkpoint signaling upon double strand breaks (DSBs), apoptosis and genotoxic stresses such as ionizing ultraviolet A light (UVA), thereby acting as a DNA damage sensor. Recognizes the substrate consensus sequence [ST]-Q. Phosphorylates 'Ser-139' of histone variant H2AX/H2AFX at double strand breaks (DSBs), thereby regulating DNA damage response mechanism. Also plays a role in pre-B cell allelic exclusion, a process leading to expression of a single immunoglobulin heavy chain allele to enforce clonality and monospecific recognition by the B-cell antigen receptor (BCR) expressed on individual B lymphocytes. After the introduction of DNA breaks by the RAG complex on one immunoglobulin allele, acts by mediating a repositioning of the second allele to pericentromeric heterochromatin, preventing accessibility to the RAG complex and recombination of the second allele. Also involved in signal transduction and cell cycle control. May function as a tumor suppressor. Necessary for activation of ABL1 and SAPK. Phosphorylates p53/TP53, FANCD2, NFKBIA, BRCA1, CTIP, nibrin (NBN), TERF1, RAD9 and DCLRE1C. May play a role in vesicle and/or protein transport. Could play a role in T-cell development, gonad and neurological function. Plays a role in replication-dependent histone mRNA degradation. Binds DNA ends.

Tissue specificity

Involvement in disease

Found in pancreas, kidney, skeletal muscle, liver, lung, placenta, brain, heart, spleen, thymus, testis, ovary, small intestine, colon and leukocytes.

Defects in ATM are the cause of ataxia telangiectasia (AT) [MIM:208900]; also known as Louis-Bar syndrome, which includes four complementation groups: A, C, D and E. This rare recessive disorder is characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. AT patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. Note=Defects in ATM contribute to T-cell acute lymphoblastic leukemia (TALL) and T-prolymphocytic leukemia (TPLL). TPLL is characterized by a high white blood cell count, with a predominance of prolymphocytes, marked splenomegaly, lymphadenopathy, skin lesions and serous effusion. The clinical course is highly aggressive, with poor response to chemotherapy and short survival time. TPLL occurs both in adults as a sporadic disease and in younger AT patients. Note=Defects in ATM contribute to B-cell non-Hodgkin lymphomas (BNHL), including mantle cell lymphoma (MCL).

Note=Defects in ATM contribute to B-cell chronic lymphocytic leukemia (BCLL). BCLL is the commonest form of leukemia in the elderly. It is characterized by the accumulation of mature CD5+ B lymphocytes, lymphadenopathy, immunodeficiency and bone marrow failure.

Sequence similarities

Belongs to the PI3/PI4-kinase family. ATM subfamily.

Contains 1 FAT domain.
Contains 1 FATC domain.
Contains 1 PI3K/PI4K domain.

Domain

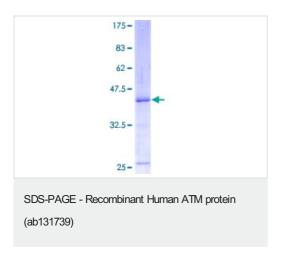
The FATC domain is required for interaction with KAT5.

Post-translational modifications

Phosphorylated by NUAK1/ARK5. Autophosphorylation on Ser-367, Ser-1893, Ser-1981 correlates with DNA damage-mediated activation of the kinase.

Acetylation, on DNA damage, is required for activation of the kinase activity, dimer-monomer transition, and subsequent autophosphorylation on Ser-1981. Acetylated in vitro by KAT5/TIP60.

Images



12.5% SDS-PAGE using ab131739 stained with Coomassie Blue.

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