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SMCHD1 Antibody

Rabbit mAb
Catalog # AP92370

Product Information

Application	WB, IHC
Primary Accession	A6NHR9
Reactivity	Human
Clonality	Monoclonal
Other Names	BAMS; FSHD2; Smchd1;
Isotype	Rabbit IgG
Host	Rabbit
Calculated MW	226374

Additional Information

Dilution	WB 1:500~1:2000 IHC 1:50~1:200
Purification	Affinity-chromatography
Immunogen	A synthesized peptide derived from human SMCHD1
Description	Required for maintenance of X inactivation in females and hypermethylation of CpG islands associated with inactive X. Involved in a pathway that mediates the methylation of a subset of CpG islands slowly and requires the methyltransferase DNMT3B (By similarity). Required for DUX4 silencing in somatic cells.
Storage Condition and Buffer	Rabbit IgG in phosphate buffered saline , pH 7.4, 150mM NaCl, 0.02% sodium azide and 50% glycerol. Store at +4°C short term. Store at -20°C long term. Avoid freeze / thaw cycle.

Protein Information

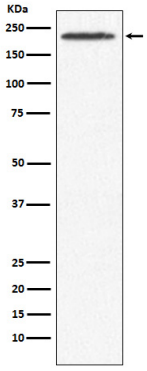
Name	SMCHD1 (HGNC:29090)
Function	Non-canonical member of the structural maintenance of chromosomes (SMC) protein family that plays a key role in epigenetic silencing by regulating chromatin architecture (By similarity). Promotes heterochromatin formation in both autosomes and chromosome X, probably by mediating the merge of chromatin compartments (By similarity). Plays a key role in chromosome X inactivation in females by promoting the spreading of heterochromatin (PubMed:23542155). Recruited to inactivated chromosome X by Xist RNA and acts by mediating the merge of chromatin compartments: promotes random chromatin interactions that span the boundaries of existing structures, leading to create a compartment-less architecture typical of inactivated chromosome X (By similarity). Required to facilitate Xist RNA spreading (By similarity). Also required for silencing of a subset of clustered autosomal loci in somatic cells, such as the DUX4 locus (PubMed:23143600). Has ATPase activity; may participate in structural manipulation of chromatin in an

ATP-dependent manner as part of its role in gene expression regulation (PubMed:29748383). Also plays a role in DNA repair: localizes to sites of DNA double-strand breaks in response to DNA damage to promote the repair of DNA double-strand breaks (PubMed:24790221, PubMed:25294876). Acts by promoting non- homologous end joining (NHEJ) and inhibiting homologous recombination (HR) repair (PubMed:25294876).

Cellular Location

Chromosome. Note=Recruited to inactivated chromosome X in females by Xist RNA (By similarity). Localizes at sites of DNA damage at double-strand breaks (DSBs) (PubMed:24790221, PubMed:25294876). {ECO:0000250|UniProtKB:Q6P5D8, ECO:0000269|PubMed:24790221, ECO:0000269|PubMed:25294876}

Images



Western blot analysis of SMCHD1 expression in 293T cell lysate.