

KD-Validated Anti-SMCHD1 Rabbit Monoclonal Antibody

Rabbit monoclonal antibody

Catalog # AGI1765

Product Information

Application	WB, ICC
Primary Accession	A6NHR9
Reactivity	Rat, Human
Clonality	Monoclonal
Isotype	Rabbit IgG
Clone Names	24GB2385
Calculated MW	226374
Gene Name	SMCHD1
Aliases	SMCHD1; Structural Maintenance Of Chromosomes Flexible Hinge Domain Containing 1; KIAA0650; FSHD2; Structural Maintenance Of Chromosomes Flexible Hinge Domain-Containing Protein 1; SMC Hinge Domain-Containing Protein 1; EC 3.6.1.-; BAMS
Immunogen	A synthesized peptide derived from human SMCHD1

Additional Information

Gene ID	23347
Other Names	Structural maintenance of chromosomes flexible hinge domain-containing protein 1, 3.6.1.-, SMCHD1 (HGNC:29090)

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}

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