



Rabbit Anti-SMCHD1 antibody

SL19929R

Product Name:	SMCHD1
Chinese Name:	SMCHD1 蛋白抗体
Alias:	KIAA0650; SMC hinge domain containing 1; SMC hinge domain containing protein 1; Smchd1; SMHD1_HUMAN; Structural maintenance of chromosomes flexible hinge domain containing 1; Structural maintenance of chromosomes flexible hinge domain-containing protein 1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Rabbit,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	226kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SMCHD1:761-860/2005
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	This gene encodes a protein which contains a hinge region domain found in members of the SMC (structural maintenance of chromosomes) protein family. [provided by RefSeq, Dec 2011]

Function:

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 de novo methyltransferase DNMT3B (By similarity). Required for DUX4 silencing in somatic cells.

Subcellular Location:

Chromosome

DISEASE:

The disease is caused by mutations affecting the gene represented in this entry. SMCHD1 mutations lead to DUX4 expression in somatic tissues, including muscle cells, when an haplotype on chromosome 4 is permissive for DUX4 expression. Ectopic expression of DUX4 in skeletal muscle activates the expression of stem cell and germline genes, and, when overexpressed in somatic cells, DUX4 can ultimately lead to cell death.

Disease description: A degenerative muscle disease characterized by slowly progressive weakness of the muscles of the face, upper-arm, and shoulder girdle. The onset of symptoms usually occurs in the first or second decade of life. Affected individuals usually present with impairment of upper extremity elevation. This tends to be followed by facial weakness, primarily involving the orbicularis oris and orbicularis oculi muscles.

SWISS:

A6NHR9

Gene ID:

23347

Database links:

[Entrez Gene: 23347](#) Human

[Entrez Gene: 74355](#) Mouse

[Omim: 614982](#) Human

[SwissProt: A6NHR9](#) Human

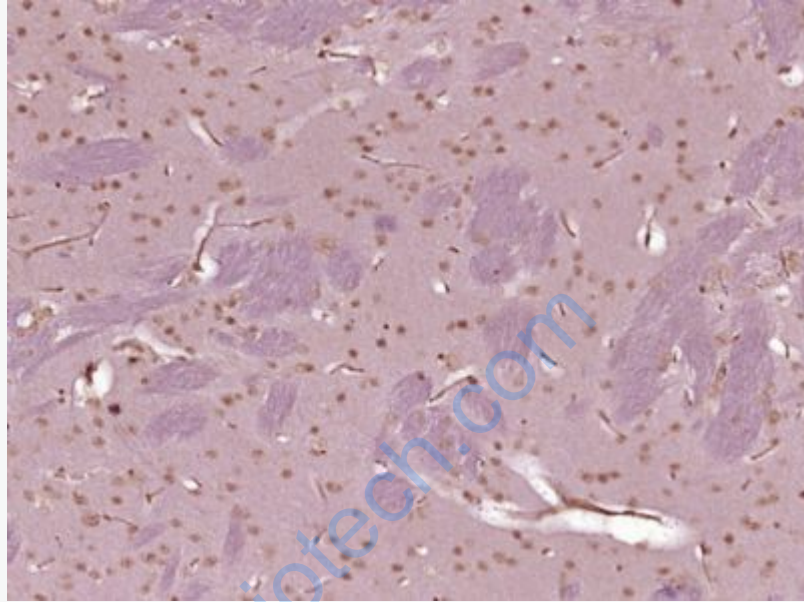
[SwissProt: Q6P5D8](#) Mouse

[Unigene: 8118](#) Human

[Unigene: 194450](#) Mouse

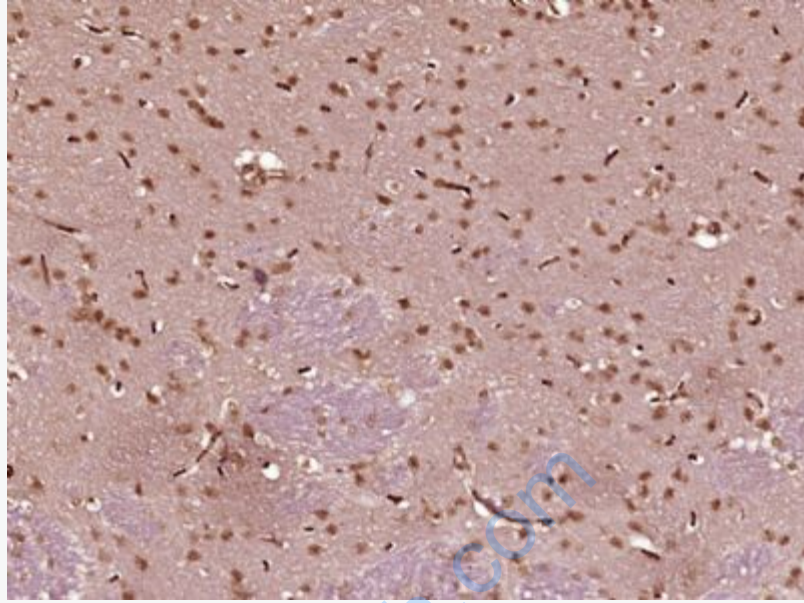
Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

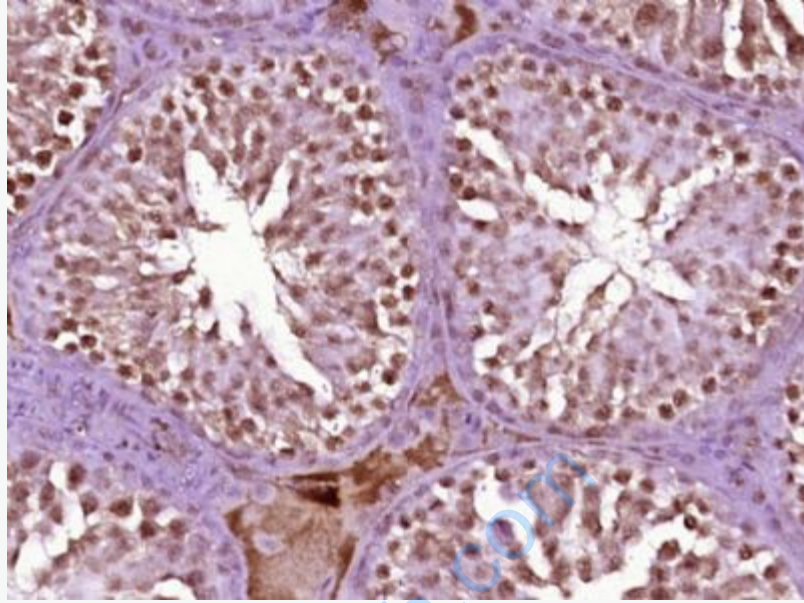


Picture:

Paraformaldehyde-fixed, paraffin embedded (Mouse brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (SMCHD1) Polyclonal Antibody, Unconjugated (SL19929R) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.



Paraformaldehyde-fixed, paraffin embedded (Rat brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (SMCHD1) Polyclonal Antibody, Unconjugated (SL19929R) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.



Paraformaldehyde-fixed, paraffin embedded (Rat testis); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (SMCHD1) Polyclonal Antibody, Unconjugated (SL19929R) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.