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Rabbit Anti-CHD7 antibody

SL13895R

Product Name:	CHD7
Chinese Name:	ATP依赖的解旋酶CHD7抗体
Alias:	ATP-dependent helicase CHD7; ATP-dependent helicase chromodomain helicase DNA binding protein 7; CHD-7; Chd7; CHD7_HUMAN; Chromodomain helicase DNA binding protein 7; chromodomain helicase DNA binding protein 7 isoform CRA_e; Chromodomain-helicase-DNA-binding protein 7; FLJ20357; FLJ20361; HH5; IS3; KAL5; KIAA1416.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,Rabbit,
Applications:	ELISA=1:500-1000IHC-P=1:400-800（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	336kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human CHD7:701-800/2997
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 that contains several helicase family domains. Mutations in this gene have been found in some patients with the CHARGE syndrome. [provided by RefSeq, Jul 2008]

Function:
Probable transcription regulator.

Subcellular Location:
Nucleus.

Tissue Specificity:
Widely expressed in fetal and adult tissues.

Post-translational modifications:
Phosphorylated upon DNA damage, probably by ATM or ATR.

DISEASE:
Defects in CHD7 are a cause of CHARGE syndrome (CHARGES) [MIM:214800]. This syndrome, which is a common cause of congenital anomalies, is characterized by a non-random pattern of congenital anomalies including choanal atresia and malformations of the heart, inner ear, and retina.
Genetic variations in CHD7 are associated with susceptibility to idiopathic scoliosis type 3 (IS3) [MIM:608765]. Idiopathic scoliosis (IS) is the most common spinal deformity in children.
Defects in CHD7 are the cause of Kallmann syndrome type 5 (KAL5) [MIM:612370]. Kallmann syndrome is a disorder that associates hypogonadotropic hypogonadism and anosmia. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In some patients other developmental anomalies can be present, which include renal agenesis, cleft lip and/or palate, selective tooth agenesis, and bimanual synkinesis. In some cases anosmia may be absent or inconspicuous.
Defects in CHD7 are a cause of idiopathic hypogonadotropic hypogonadism (IHH) [MIM:146110]. IHH is defined as a deficiency of the pituitary secretion of follicle-stimulating hormone and luteinizing hormone, which results in the impairment of pubertal maturation and of reproductive function.

Similarity:
Belongs to the SNF2/RAD54 helicase family.
Contains 2 chromo domains.
Contains 1 helicase ATP-binding domain.
Contains 1 helicase C-terminal domain.

SWISS:
Q9P2D1

Gene ID:
55636

Database links:

	Entrez Gene: 55636 Human Entrez Gene: 320790 Mouse Omim: 608892 Human SwissProt: Q9P2D1 Human SwissProt: A2AJK6 Mouse Unigene: 20395 Human Unigene: 138792 Mouse **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.