

CHD7 rabbit pAb

Catalog No: YT7184

Reactivity: Human; Mouse

Applications: IHC;IF

Target: CHD7

Gene Name: CHD7 KIAA1416

Q9P2D1

A2AJK6

Protein Name: CHD7

Human Gene Id: 55636

Human Swiss Prot

No:

Mouse Gene ld: 320790

Mouse Swiss Prot

No:

Immunogen: Synthesized peptide derived from human CHD7 AA range: 1703-1753

Specificity: This antibody detects endogenous levels of CHD7 at Human/Mouse

Formulation: Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Source: Polyclonal, Rabbit, IgG

Dilution: IHC 1 ? 50-200. IF 1:50-200

Purification: The antibody was affinity-purified from rabbit antiserum by affinity-

chromatography using epitope-specific immunogen.

Concentration: 1 mg/ml

Storage Stability: -15°C to -25°C/1 year(Do not lower than -25°C)

1/2



Molecularweight: 330kD

Background: 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. Two transcript variants encoding different isoforms have been found

for this gene. [provided by RefSeq, Oct 2015],

Function: disease:Defects in CHD7 are a cause of CHARGE syndrome [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., disease: 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., disease: 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

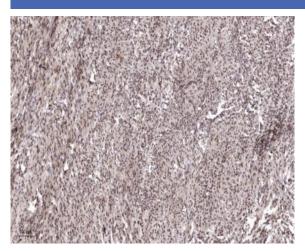
olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropi

Subcellular Location :

[Isoform 1]: Nucleus .; [Isoform 3]: Nucleus, nucleolus .

Expression: Widely expressed in fetal and adult tissues.

Products Images



Immunohistochemical analysis of paraffin-embedded human Colon cancer. 1, Antibody was diluted at 1:200(4° overnight). 2, Tris-EDTA,pH9.0 was used for antigen retrieval. 3,Secondary antibody was diluted at 1:200(room temperature, 45min).