abcam

Product datasheet

Mouse Chd7 peptide ab31859

Description

Product name	Mouse Chd7 peptide
Animal free	No
Nature	Synthetic
Species	Mouse

Specifications

Our Abpromise guarantee covers the use of **ab31859** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Form	Liquid
Additional notes	*- First try to dissolve a small amount of peptide in either water or buffer. The more charged residues on a peptide, the more soluble it is in aqueous solutions.* *- If the peptide doesn't dissolve try an organic solvent e.g. DMSO, then dilute using water or buffer.* *- Consider that any solvent used must be compatible with your assay. If a peptide does not dissolve and you need to recover it, lyophilise to remove the solvent.* *- Gentle warming and sonication can effectively aid peptide solubilisation. If the solution is cloudy or has gelled the peptide may be in suspension rather than solubilised.* *- Peptides containing cysteine are easily oxidised, so should be prepared in solution just prior to use.*

Preparation and Storage

Stability and Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request.

General Info

Function	Probable transcription regulator.
Tissue specificity	Widely expressed in fetal and adult tissues.
Involvement in 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.
Sequence similarities	Belongs to the SNF2/RAD54 helicase family. Contains 2 chromo domains. Contains 1 helicase ATP-binding domain. Contains 1 helicase C-terminal domain.
Post-translational modifications	Phosphorylated upon DNA damage, probably by ATM or ATR.
Cellular localization	Nucleus.

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