

SIX5 Rabbit pAb

Catalog # AP99449

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q8N196
Other Accession	Q8N196
Predicted	Rat, Human, Mouse, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	74562
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SIX5
Epitope Specificity	221-320/739
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the SIX/Sine oculis homeobox family. Contains 1 homeobox DNA-binding domain.
DISEASE	Defects in SIX5 are the cause of branchiootorenal syndrome type 2 (BOR2) [MIM:610896]. BOR is an autosomal dominant disorder manifested by various combinations of preauricular pits, branchial fistulae or cysts, lacrimal duct stenosis, hearing loss, structural defects of the outer, middle, or inner ear, and renal dysplasia. Associated defects include asthenic habitus, long narrow facies, constricted palate, deep overbite, and myopia. Hearing loss may be due to mondini type cochlear defect and stapes fixation. Penetrance of BOR syndrome is high, although expressivity can be extremely variable.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene is a homeodomain-containing transcription factor that appears to function in the regulation of organogenesis. This gene is located downstream of the dystrophia myotonica-protein kinase gene. Mutations in this gene are a cause of branchiootorenal syndrome type 2. [provided by RefSeq, Jul 2009]

Additional Information

Gene ID	147912
Other Names	BOR2; DMAHP; MDMAHP; TrexBF; SIX5_HUMAN; SIX5; DM locus-associated homeodomain protein; Sine oculis homeobox homolog 5; SIX5_MOUSE; DM locus-associated homeodomain protein homolog; SIX homeobox 5; sine oculis homeobox (Drosophila) homolog 5; sine oculis homeobox homolog 5 (Drosophila)
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500

Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.
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Protein Information

Name	SIX5
Synonyms	DMAHP
Function	Transcription factor that is thought to be involved in regulation of organogenesis. May be involved in determination and maintenance of retina formation. Binds a 5'-GGTGTCTAG-3' motif present in the ARE regulatory element of ATP1A1. Binds a 5'-TCA[AG][AG]TTNC-3' motif present in the MEF3 element in the myogenin promoter, and in the IGFBP5 promoter (By similarity). Thought to be regulated by association with Dach and Eya proteins, and seems to be coactivated by EYA1, EYA2 and EYA3 (By similarity).
Cellular Location	Cytoplasm. Nucleus {ECO:0000255 PROSITE-ProRule:PRU00108, ECO:0000269 PubMed:12500905}
Tissue Location	Expressed in adult but not in fetal eyes. Found in corneal epithelium and endothelium, lens epithelium, ciliary body epithelia, cellular layers of the retina and the sclera

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