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SIX1 Rabbit pAb

Catalog # AP99448

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q15475
Other Accession	Q15475
Predicted	Rat, Human, Mouse, Rabbit, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	32210
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SIX1
Epitope Specificity	151-250/284
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 SIX1 are the cause of deafness autosomal dominant type 23 (DFNA23) [MIM:605192]. A form of non-syndromic deafness characterized by prelingual, bilateral, symmetric hearing loss with a conductive component present in some but not all patients. Defects in SIX1 are the cause of branchiootic syndrome type 3 (BOS3) [MIM:608389]. BOS3 is a syndrome characterized by usually bilateral branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, and structural defects of the outer, middle or inner ear. Otic defects include malformed and hypoplastic pinnae, a narrowed external ear canal, bulbous internal auditory canal, stapes fixation, malformed and hypoplastic cochlea. Branchial and otic anomalies are as those seen in individuals with the branchiootorenal syndrome. However, renal anomalies are absent in branchiootic syndrome patients. Note=Defects in SIX1 could be a cause of branchiootorenal syndrome (BOR). 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 homeobox protein that is similar to the Drosophila 'sine oculis' gene product. This gene is found in a cluster of related genes on chromosome 14 and is thought to be involved in limb development. Defects in this gene are a cause of autosomal dominant deafness type 23 (DFNA23) and branchiootic syndrome type 3 (BOS3). [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	6495
Other Names	BOS3; DFNA23; TIP39; SIX1_HUMAN; SIX1; Sine oculis homeobox homolog 1; SIX1_MOUSE; Q8BSP4_MOUSE; SIX homeobox 1; sine oculis homeobox (Drosophila) homolog 1; sine oculis homeobox homolog 1 (Drosophila); deafness, autosomal dominant 23
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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.

Protein Information

Name	SIX1
Function	Transcription factor that is involved in the regulation of cell proliferation, apoptosis and embryonic development (By similarity). Plays an important role in the development of several organs, including kidney, muscle and inner ear (By similarity). Depending on context, functions as a transcriptional repressor or activator (By similarity). Lacks an activation domain, and requires interaction with EYA family members for transcription activation (PubMed:15141091). Mediates nuclear translocation of EYA1 and EYA2 (PubMed:19497856). Binds the 5'-TCA[AG][AG]TTNC-3' motif present in the MEF3 element in the MYOG promoter and CIDEA enhancer (PubMed:15141091, PubMed:19497856, PubMed:23435380, PubMed:27923061). Regulates the expression of numerous genes, including MYC, CCND1 and EZR (By similarity). Acts as an activator of the IGFBP5 promoter, probably coactivated by EYA2 (By similarity). Repression of precursor cell proliferation in myoblasts is switched to activation through recruitment of EYA3 to the SIX1-DACH1 complex (By similarity). During myogenesis, seems to act together with EYA2 and DACH2 (By similarity). Regulates the expression of CCNA1 (PubMed:15123840). Promotes brown adipocyte differentiation (By similarity).
Cellular Location	Nucleus. Cytoplasm
Tissue Location	Specifically expressed in skeletal muscle.

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