

Rabbit Anti-SIX1/AP Conjugated antibody

SL17504R-AP

Product Name	Anti-SIX1/AP
Chinese Name	碱性磷酸酶 (AP) 标记的同源盒蛋白 SIX1 抗体
Alias	BOS3; DFNA23; Homeobox protein SIX1; OTTHUMP00000179042; Sine oculis homeobox homolog 1; SIX homeobox 1; SIX1; SIX1_HUMAN; TIP39.
Research Area	Cell biology Developmental biology transcriptional regulatory factor Epigenetics
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	(predicted:Human,Mouse,Rat,Cow,Rabbit,Sheep) WB=1000-10000,IHC-P=1:100-500,IHC-F=1:100-500,ELISA=1:500-5000
Applications	not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	32kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human SIX1
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol. 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 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Storage	
Product Detail	background: 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,

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Function:

May be involved in limb tendon and ligament development.

Subcellular Location:

Nucleus.

Tissue Specificity:

Specifically expressed in skeletal muscle.

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.

Similarity:

Belongs to the SIX/Sine oculis homeobox family.
Contains 1 homeobox DNA-binding domain.

Database links:

[Entrez Gene: 6495](#) Human

[Entrez Gene: 20471](#) Mouse

[Entrez Gene: 114634](#) Rat

[Omim: 601205](#) Human

[SwissProt: Q15475](#) Human

[SwissProt: Q62231](#) Mouse

[SwissProt: Q8BSP4](#) Mouse

[Unigene: 54416](#) Human

[Unigene: 713114](#) Human

[Unigene: 4645](#) Mouse

[Unigene: 23396](#) Rat

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.