

# Rabbit Anti-SIX5 antibody

# SL17505R

<b>Product Name:</b>	SIX5
Chinese Name:	Diabetes相关同源结构域蛋白SIX5抗体
Alias:	BOR2; DM locus associated homeodomain protein; DM locus-associated homeodomain protein; DMAHP; Dystrophia myotonica associated homeodomain protein; Homeobox protein SIX5; Sine oculis homeobox homolog 5; sine oculis related homeobox 5 homolog (Drosophila); SIX homeobox 5; SIX5; SIX5_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Dog, Cow, Horse, Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	75kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	lmg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SIX5:221-320/739
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	<u>PubMed</u>
Product Detail:	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]

#### **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'-GGTGTCAG-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.

#### **Subcellular Location:**

Cytoplasm. Nucleus.

### **Tissue Specificity:**

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.

#### 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.

#### Similarity:

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

## SWISS:

O8N196

#### Gene ID:

147912

## Database links:

Entrez Gene: 147912 Human

Entrez Gene: 20475 Mouse

Omim: 600963 Human

SwissProt: Q8N196 Human

SwissProt: P70178 Mouse

Unigene: 43314 Human

Unigene: 635370 Human

Unigene: 3410 Mouse

# Important Note:

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