

Rabbit Anti-EYA1 antibody

SL20158R

Product Name:	EYA1
Chinese Name:	转录 因子 EYA1 抗体
Alias:	BOP; BOR; Eya1; EYA1_HUMAN; eyes absent 1; eyes absent 1 homolog; eyes absent homolog 1 (Drosophila); Eyes absent homolog 1; eyes absent homolog 1; MGC141875
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,
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:	65kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EYA1:231-330/592
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	Preservative: 15mM Sodium Azide, Constituents: 1% BSA, 0.01M PBS, pH 7.4
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:	PubMed
Product Detail:	A gene on chromosome 8q13.3 encodes EYA1 (eyes absent), a protein with 16 exons. EYA1 is one of four members of the eyes absent family. A 271 amino acid domain at the carboxyl terminal is highly conserved amongst the members of the eyes absent family, while the PST (proline-serine-threonin)-rich amino terminal is highly divergent. EYA is expressed in flexor tendons and the developing central nervous system, kidney, eye and ear. EYA1 acts a transcriptional activator in connective tissue patterning through its PST

domain, which functions as a transactivation domain. EYA1 plays a critical role in the development of the inner ear and kidney. EYA is involved in early inductive signaling, acting upstream of GDNF. EYA1 has been implicated in the autosomal dominant disorders branchio-oto-renal (BOR) syndrome and branhio-oto (BO) syndrome.

Function:

Tyrosine phosphatase that specifically dephosphorylates 'Tyr-142' of histone H2AX (H2AXY142ph). 'Tyr-142' phosphorylation of histone H2AX plays a central role in DNA repair and acts as a mark that distinguishes between apoptotic and repair responses to genotoxic stress. Promotes efficient DNA repair by dephosphorylating H2AX, promoting the recruitment of DNA repair complexes containing MDC1. Its function as histone phosphatase probably explains its role in transcription regulation during organogenesis. Seems to coactivate SIX2, SIX4 and SIX5. May be required for normal development of branchial arches, ear and kidney.

Subunit:

Probably interacts with SIX2, SIX4 and SIX5.

Subcellular Location:

Cytoplasm. Nucleus. Localizes at sites of DNA damage at double-strand breaks.

Tissue Specificity:

In the embryo, highly expressed in kidney with lower levels in brain. Weakly expressed in lung. In the adult, highly expressed in heart and skeletal muscle. Weakly expressed in brain and liver. No expression in eye or kidney.

Post-translational modifications:

Sumoylated by SUMO1.

DISEASE:

Defects in EYA1 are the cause of branchiootorenal syndrome type 1 (BOR1) [MIM:113650]; also known as Melnick-Fraser syndrome. 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.

Defects in EYA1 are the cause of otofaciocervical syndrome (OFCS) [MIM:166780]. The syndrome is characterized by trophic alterations of the facies and shoulder girdle in addition to the malformations seen in BOR.

Defects in EYA1 are the cause of branchiootic syndrome type 1 (BOS1) [MIM:602588]; also known as BO syndrome type 1 or branchiootic dysplasia. Individuals with BOS1 are affected by the same branchial and otic anomalies as those seen in individuals with BOR1, but lack renal anomalies.

Similarity: Belongs to the HAD-like hydrolase superfamily. EYA family.
SWISS: Q99502
Gene ID: 2138
Database links:
Entrez Gene: 395718Chicken
Entrez Gene: 511188Cow
Entrez Gene: 477910Dog
Entrez Gene: 2138Human
Entrez Gene: 14048Mouse
Entrez Gene: 502935Rat
Omim: 601653Human
SwissProt: Q9YHA0Chicken
SwissProt: Q99502Human
SwissProt: P97767Mouse
<u>Unigene: 491997</u> Human
Unigene: 250185Mouse
N.
Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

