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EYA1 Rabbit pAb

Catalog # AP96555

Product Information

Application	WB
Primary Accession	Q99502
Other Accession	Q99502
Reactivity	Human
Host	Rabbit
Clonality	Polyclonal
Calculated MW	64593
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human EYA1
Epitope Specificity	21-120/592
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 HAD-like hydrolase superfamily. EYA family.
SUBUNIT	Probably interacts with SIX2, SIX4 and SIX5.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	2138
Other Names	BOP; BOR; BOS1; OFC1; OTFCS; EYA1_CHICK; EYA1; Eyes absent homolog 1; 3.1.3.16; 3.1.3.48; EYA1_HUMAN; EYA1_MOUSE; EYA transcriptional coactivator and phosphatase 1; eyes absent (Drosophila) homolog 1; eyes absent homolog 1 (Drosophila)
Dilution	WB=1:500-2000
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	EYA1
Function	Functions both as protein phosphatase and as transcriptional coactivator for SIX1, and probably also for SIX2, SIX4 and SIX5 (By similarity). Tyrosine phosphatase that dephosphorylates 'Tyr-142' of histone H2AX (H2AXY142ph) and promotes efficient DNA repair via the recruitment of DNA repair complexes containing MDC1. '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 (PubMed:19234442). Its function as histone phosphatase may contribute to its function in transcription regulation during organogenesis (By similarity). Also has phosphatase activity with proteins phosphorylated on Ser and Thr residues (in vitro) (By similarity). Required for normal embryonic development of the craniofacial and trunk skeleton, kidneys and ears (By similarity). Together with SIX1, it plays an important role in hypaxial muscle development; in this it is functionally redundant with EYA2 (By similarity).
Cellular Location	Cytoplasm. Nucleus Note=Localizes at sites of DNA damage at double-strand breaks (DSBs)
Tissue Location	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

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