

Rabbit Anti-EYA4 antibody

SL13126R

Product Name:	EYA4
Chinese Name:	EYA4蛋白抗体
Alias:	CMD1J; Deafness, autosomal dominant 10; DFNA 10; DFNA10; dJ78N10.1 (eyes absent (Drosophila) homolog 4); dJ78N10.1 (eyes absent); EYA 4; eya4; EYA4_HUMAN; Eyes absent 4; Eyes absent homolog 4 (Drosophila); Eyes absent homolog 4; HGNC:3522; OTTHUMP00000040267.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,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:	70kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EYA4:301-400/639
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:	PubMed
Product Detail:	A gene of chromosome 6q23 encodes the 640 amino acid protein, EYA4 (eyes absent) (1). EYA is one of four members of the eyes absent family (1). A 271 amino acid domain at the carboxyl terminal is highly conserved amongst the members of the eyes absent family (1). EYA4 is expressed in the craniofacial mesenchyme, the

dermamyotome, and the limb (1). The conserved region in other EYA proteins interacts with SIX, DACH, and G-proteins, which regulate transcription in early embryonic development (1,2,3,4). SIX translocates EYA1-3 to the nucleus, and G-proteins can stop this interaction (3,4). Premature stop codon mutations in EYA4 cause postlingual, progressive autosomal dominant hearing loss in humans (2). This shows that EYA4 is also vital to the mature organ of Corti (2). EYA4 may cause oculo-dento-digital syndrome, based on its expression pattern and map postion (1).

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. May be involved in development of the eye.

Subcellular Location: Cytoplasm. Nucleus.

Tissue Specificity: Highly expressed in heart and skeletal muscle.

DISEASE:

Defects in EYA4 are the cause of deafness autosomal dominant type 10 (DFNA10) [MIM:601316]. DFNA10 is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. Defects in EYA4 are the cause of cardiomyopathy dilated type 1J (CMD1J) [MIM:605362]. Dilated cardiomyopathy is a disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.

Similarity:

Belongs to the HAD-like hydrolase superfamily. EYA family.

SWISS:

095677

Gene ID: 2070

Database links:

Entrez Gene: 2070Human

Entrez Gene: 14051 Mouse
Omim: 603550Human
SwissProt: 095677Human
SwissProt: Q9Z191Mouse
Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications
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