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PHOX2A Rabbit pAb

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Catalog # AP54553

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	O14813
Predicted	Rat, Human, Mouse, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	29653
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PHOX2A
Epitope Specificity	41-140/284
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Nucleus.
DISEASE	Defects in PHOX2A are the cause of congenital fibrosis of extraocular muscles type 2 (CFEOM2) [MIM:602078]. CFEOM encompasses several different inherited strabismus syndromes characterized by congenital restrictive ophthalmoplegia affecting extraocular muscles innervated by the oculomotor and/or trochlear nerves. CFEOM is characterized clinically by anchoring of the eyes in downward gaze, ptosis, and backward tilt of the head. CFEOM2 may result from the aberrant development of the oculomotor (nIII), trochlear (nIV) and abducens (nVI) cranial nerve nuclei.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene contains a paired-like homeodomain most similar to that of the Drosophila aristaless gene product. The encoded protein plays a central role in development of the autonomic nervous system. It regulates the expression of tyrosine hydroxylase and dopamine beta-hydroxylase, two catecholaminergic biosynthetic enzymes essential for the differentiation and maintenance of the noradrenergic neurotransmitter phenotype. The encoded protein has also been shown to regulate transcription of the alpha3 nicotinic acetylcholine receptor gene. Mutations in this gene have been associated with autosomal recessive congenital fibrosis of the extraocular muscles. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	401
Other Names	Paired mesoderm homeobox protein 2A, ARIX1 homeodomain protein, Aristaless homeobox protein homolog, Paired-like homeobox 2A, PHOX2A, ARIX, PMX2A

Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	PHOX2A
Synonyms	ARIX, PMX2A
Function	May be involved in regulating the specificity of expression of the catecholamine biosynthetic genes. Involved in the development of several major noradrenergic neuron populations, including the locus coeruleus. Acts as a transcription activator/factor. Could maintain the noradrenergic phenotype (By similarity).
Cellular Location	Nucleus {ECO:0000255|PROSITE-ProRule:PRU00108}.

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.