

## PHOX2A Polyclonal Antibody

Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP54553

## **Product Information**

**Application** WB, IHC-P, IHC-F, IF, ICC, E

**Primary Accession** 014813 Reactivity Rat, Bovine Host Rabbit Clonality Polyclonal Calculated MW 29653 **Physical State** Liquid

**Immunogen** KLH conjugated synthetic peptide derived from human PHOX2A

41-140/284 **Epitope Specificity** 

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

**SIMILARITY** 

Belongs to the paired homeobox family. Contains 1 homeobox DNA-binding

domain.

**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.

This product as supplied is intended for research use only, not for use in **Important Note** 

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=1:100-500,IF=1:100-50

0,ELISA=1:5000-10000

Format 0.01M TBS(pH7.4) with 1% BSA, 0.09% (W/V) sodium azide and 50% Glyce

**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. Acts as a transcription activator/factor.

Could maintain the noradrenergic phenotype.

**Cellular Location** Nucleus {ECO:0000255 | PROSITE-ProRule:PRU00108}.

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