

CRX1 Rabbit pAb

Catalog # AP99046

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	O43186
Other Accession	O43186
Host	Rabbit
Clonality	Polyclonal
Calculated MW	32261
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human CRX
Epitope Specificity	51-150/299
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 paired homeobox family. Contains 1 homeobox DNA-binding domain.
SUBUNIT	Interacts with SCA7. Interacts with RAX2. Interacts (via the homeobox) with NRL (via the leucine-zipper domain). Interacts with PDC.
DISEASE	Defects in CRX are the cause of Leber congenital amaurosis type 7 (LCA7) [MIM:613829]. LCA designates a clinically and genetically heterogeneous group of childhood retinal degenerations, generally inherited in an autosomal recessive manner. Affected infants have little or no retinal photoreceptor function as tested by electroretinography. LCA represents the most common genetic cause of congenital visual impairment in infants and children. [DISEASE] Defects in CRX are the cause of cone-rod dystrophy type 2 (CORD2) [MIM:120970]; also known as cone-rod retinal dystrophy 2 (CRD2). CORDs are inherited retinal dystrophies belonging to the group of pigmentary retinopathies. CORDs are characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. Defects in CRX are a cause of retinitis pigmentosa (RP) [MIM:268000]. RP leads to degeneration of retinal photoreceptor cells. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well.
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 is a photoreceptor-specific transcription factor which plays a role in the differentiation of photoreceptor cells. This homeodomain protein is necessary for the maintenance of normal cone and rod function. Mutations in this gene are associated with photoreceptor degeneration, Leber congenital amaurosis type III and the autosomal dominant cone-rod dystrophy 2. Several alternatively spliced transcript variants of this gene have been described, but the full-length nature of some variants has not been determined. [provided by RefSeq, Jul 2008].

Additional Information

Gene ID	1406
Other Names	CORD2; CRD; LCA7; OTX3; Crx1; CRX_BOVIN; CRX; CRX_CANLF; CRX_HUMAN; CRX_MOUSE; cone-rod homeobox; orthodenticle homeobox 3
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	CRX
Synonyms	CORD2
Function	Transcription factor that binds and transactivates the sequence 5'-TAATC[CA]-3' which is found upstream of several photoreceptor-specific genes, including the opsin genes. Acts synergistically with other transcription factors, such as NRL, RORB and RAX, to regulate photoreceptor cell-specific gene transcription. Essential for the maintenance of mammalian photoreceptors.
Cellular Location	Nucleus {ECO:0000255 PROSITE-ProRule:PRU00108}.
Tissue Location	Retina.

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