

Recombinant Human PAX6 protein ab134885

Description

Product name	Recombinant Human PAX6 protein
Purity	> 90 % SDS-PAGE. ab134885 was expressed in E. coli as inclusion bodies, refolded using “temperature shift inclusion body refolding” technology, chromatographically purified and sterile filtered.
Expression system	Escherichia coli
Accession	<u>P26367</u>
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	29aa_Tag_QNSHSGVNQLGGVFVNGRPLPDSTRQKMVELA HSGARPCDI SRILQVSNGCVSKILGRYYETGSIRPRAIGGSKPRVATPEVV SKIAQYKR ECPSIFAWEIRDRLLESEGVTNDNIPSVSSINRVLRNLASE KQQMGADGM YDKLRMLNGQTGSWGTRPGWYPGTSVPGQPTQDGCQQ QEGGGENTNSISS NGEDSDEAQMRLQLKRKLQRNRTSFTQEIEALEKEFER THYPDVFAER LAAKIDLPEARIQVWFNSRRRAKWRREEKLRNQRRQASNT PSHIPSSSFS TSVYQPIQPPTPVSSFTSGSMLGRTDTALTNTYSALPPMP SFTMANNLP MQPPVPSQTSSYSCLPTSPSVNGRSYDTYTPPHMQTHM NSQPMGTSGTT STGLISPGVSPVQVPGSEPDMSQYWPRQLQEESGGGG SPGRRRRRRRRRR RR
Predicted molecular weight	49 kDa including tags
Amino acids	2 to 422

Specifications

Our **Abpromise guarantee** covers the use of **ab134885** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Functional Studies
	SDS-PAGE
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles. pH: 8.00 Constituents: Potassium chloride, 0.05% DTT, 0.32% Tris HCl, 0.02% EDTA, Glycerol, Sodium chloride, 3.4% DL-Arginine
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General Info

Function	Transcription factor with important functions in the development of the eye, nose, central nervous system and pancreas. Required for the differentiation of pancreatic islet alpha cells (By similarity). Competes with PAX4 in binding to a common element in the glucagon, insulin and somatostatin promoters. Regulates specification of the ventral neuron subtypes by establishing the correct progenitor domains (By similarity). Isoform 5a appears to function as a molecular switch that specifies target genes.
Tissue specificity	Fetal eye, brain, spinal cord and olfactory epithelium. Isoform 5a is less abundant than the PAX6 shorter form.
Involvement in disease	<p>Defects in PAX6 are the cause of aniridia (AN) [MIM:106210]. A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time.</p> <p>Defects in PAX6 are a cause of Peters anomaly (PAN) [MIM:604229]. Peters anomaly consists of a central corneal leukoma, absence of the posterior corneal stroma and Descemet membrane, and a variable degree of iris and lenticular attachments to the central aspect of the posterior cornea.</p> <p>Defects in PAX6 are a cause of foveal hypoplasia (FOVHYP) [MIM:136520]. Foveal hypoplasia can be isolated or associated with presenile cataract. Inheritance is autosomal dominant.</p> <p>Defects in PAX6 are a cause of keratitis hereditary (KERH) [MIM:148190]. An ocular disorder characterized by corneal opacification, recurrent stromal keratitis and vascularization.</p> <p>Defects in PAX6 are a cause of coloboma ocular (COLO) [MIM:120200]; also known as uveoretinal coloboma or coloboma of iris, choroid and retina. Ocular colobomas are a set of malformations resulting from abnormal morphogenesis of the optic cup and stalk, and the fusion of the fetal fissure (optic fissure). Severe colobomatous malformations may cause as much as 10% of the childhood blindness. The clinical presentation of ocular coloboma is variable. Some individuals may present with minimal defects in the anterior iris leaf without other ocular defects. More complex malformations create a combination of iris, uveoretinal and/or optic nerve defects without or with microphthalmia or even anophthalmia.</p> <p>Defects in PAX6 are a cause of coloboma of optic nerve (COLON) [MIM:120430].</p> <p>Defects in PAX6 are a cause of bilateral optic nerve hypoplasia (BONH) [MIM:165550]; also known as bilateral optic nerve aplasia. A congenital anomaly in which the optic disc appears abnormally small. It may be an isolated finding or part of a spectrum of anatomic and functional</p>

abnormalities that includes partial or complete agenesis of the septum pellucidum, other midline brain defects, cerebral anomalies, pituitary dysfunction, and structural abnormalities of the pituitary.

Defects in PAX6 are a cause of aniridia cerebellar ataxia and mental deficiency (ACAMD) [MIM:206700]; also known as Gillespie syndrome. A rare condition consisting of partial rudimentary iris, cerebellar impairment of the ability to perform coordinated voluntary movements, and mental retardation.

Sequence similarities

Belongs to the paired homeobox family.

Contains 1 homeobox DNA-binding domain.

Contains 1 paired domain.

Developmental stage

Expressed in the developing eye and brain.

**Post-translational
modifications**

Ubiquitinated by TRIM11, leading to ubiquitination and proteasomal degradation.

Cellular localization

Nucleus.

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