G-Biosciences Image not found or type unknown

Immunotag™ Pax-6 Monoclonal Antibody

Antibody Specification

Catalog No.	ITM0508
Product Description	Immunotag™ Pax-6 Monoclonal Antibody
Size	50 µg, 100 µg
Conjugation	HRP, Biotin, FITC, Alexa Fluor® 350, Alexa Fluor® 405, Alexa Fluor® 488, Alexa Fluor® 555, Alexa Fluor® 594, Alexa Fluor® 647
IMPORTANT NOTE	This product is custom manufactured with a lead time of 3-4 weeks. Once in production, this item cannot be cancelled from an order and is not eligible for return.
Target Protein	Pax-6
Clonality	Monoclonal
Storage/Stability	-20°C/1 year
Application	WB,FCM,ELISA
Recommended Dilution	Western Blot: 1/500 - 1/2000. Flow cytometry: 1/200 - 1/400. ELISA: 1/10000. Not yet tested in other applications.
Concentration	1 mg/ml
Reactive Species	Human
Host Species	Mouse
Immunogen	Purified recombinant fragment of human Pax-6 expressed in E. Coli.
Specificity	Pax-6 Monoclonal Antibody detects endogenous levels of Pax-6 protein.
Purification	Affinity purification
Form	Ascitic fluid containing 0.03% sodium azide.
Gene Name	PAX6
Accession No.	P26367 P63015
Alternate Names	PAX6; AN2; Paired box protein Pax-6; Aniridia type II protein; Oculorhombin

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Description	paired box 6(PAX6) Homo sapiens This gene encodes a homeobox and paired domain-containing protein that binds DNA and functions as a regulator of transcription. Activity of this protein is key in the development of neural tissues, particularly the eye. This gene is regulated by multiple enhancers located up to hundreds of kilobases distant from this locus. Mutations in this gene or in the enhancer regions can cause ocular disorders such as aniridia and Peter's anomaly. Use of alternate promoters and alternative splicing result in multiple transcript variants encoding different isoforms. [provided by RefSeq, Jul 2015],
Cell Pathway/ Category	Maturity onset diabetes of the young,
Protein Expression	Brain,Cerebellum,Lung,
Subcellular Localization	nuclear chromatin,nucleus,nucleoplasm,cytoplasm,

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Protein Function	developmental stage:Expressed in the developing eye and brain.,disease:Defects in PAX6 are a cause of autosomal dominant keratitis [MIM:148190]. It is an eye disorder characterized by corneal opacification and vascularization, and by foveal hypoplasia.,disease:Defects in PAX6 are a cause of bilateral optic nerve hypoplasia [MIM:165550]; also known as bilateral optic nerve aplasia. Inheritance is autosomal dominant.,disease:Defects in PAX6 are a cause of coloboma of optic nerve [MIM:120430].,disease:Defects in PAX6 are a cause of ectopia pupillae [MIM:129750]. It is a congenital eye malformation in which the pupils are displaced from their normal central position.,disease:Defects in PAX6 are a cause of foveal hypoplasia [MIM:136520]. Foveal hypoplasia can be isolated or associated with presenile cataract. Inheritance is autosomal dominant.,disease:Defects in PAX6 are a cause of Gillespie syndrome [MIM:206700]; also called aniridia cerebellar ataxia and mental deficiency. Gillespie syndrome is a rare condition consisting of partial rudimentary iris, cerebellar impairment of the ability to perform smoothly coordinated voluntary movements, and mental retardation. It is not yet clear whether the disorder has an autosomal recessive or dominant inheritance.,disease:Defects in PAX6 are a cause of ocular coloboma [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.,disease:Defects in PAX6 are a cause of Peters anomaly [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.,disease:Defects in PAX6 are the cause of aniridia type II (AN2) [MIM:106210]. AN2 is a bilateral panocular disorder characterized by complete or partial absence of the iris, absence of the fovea and malformations of the lens and anterior chamber. Severe age-related corneal degeneration is a frequent complication which contributes to a poor visual prognostis in aniridia. About one third of the cases are sporadic, and two thirds are familial, with autosomal dominant inheritance and high penetrance. Nearly one third of sporadic AN patients develop Wilms tumor in association with genitourinary anomalies and mental retardation (WAGR syndrome) as a consequence of heterozygous (sub)microscopic deletions of chromosome 11p13.,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.,similarity:Belongs to the paired homeobox family.,similarity:Contains 1 homeobox DNA-binding domain.,similarity:Contains 1 paired domain.,subunit:Interacts with MAF and MAFB.,tissue specificity:Fetal eye, brain, spinal cord and olfactory epithelium. Isoform 5a is less abundant than the PAX6 shorter form.,
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