PAX6 Rabbit pAb

CatalogNo: YT7954

Key Features

Host Species
- Rabbit

Reactivity
- Human,Rat,Mouse,

Applications
- WB,ELISA

MW
- 46kD (Calculated)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:1000-2000
ELISA 1:5000-20000

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

Immunogen	Synthesized peptide derived from human PAX6
Specificity	This antibody detects endogenous levels of Human PAX6

Target Information

Gene name	PAX6 AN2

Protein Name Paired box protein Pax-6

Organism	Gene ID	UniProt ID
Human	5080;	P26367;
Mouse	18508;	P63015;
Rat	25509;	P63016;

Cellular Localization Nucleus .; [Isoform 1]: Nucleus .; [Isoform 5a]: Nucleus .

Tissue specificity [Isoform 1]: Expressed in lymphoblasts. ; [Isoform 5a]: Weakly expressed in lymphoblasts.

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.,

Validation Data

Contact information

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Please scan the QR code to access additional product information:
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