

BACKGROUND

Several gene families have roles in regulating developmental programs. One such family, called PAX genes, derives its name from a conserved DNA sequence motif called the paired box, a conserved 128 amino acid domain in the amino-terminal portion of the protein. The PAX genes are a relatively small family of developmental genes that are grouped into four classes on the basis of their structural similarity, depending on sequence homology, the presence or absence of an octapeptide domain, and either a homeodomain or partial homeodomain. The paired box and homeodomains encode DNA binding domains within the PAX proteins, so each protein is able to act as a transcription factor regulating the expression of a range of downstream genes. An additional domain in the PAX genes is the transactivation domain within the carboxyl terminus of each PAX protein, which is a serine- and threonine-rich domain responsible for transcriptional activation of target genes. PAX genes have been identified in a wide variety of species. Nine PAX genes, PAX1-PAX9, have been described in humans and mice. The expression of PAX genes in many tissues during embryogenesis is associated with critical roles for these genes during development. In particular, mutations in four of the PAX genes, PAX2, PAX3, PAX6 and PAX8, have been shown to cause human developmental disorders. A number of PAX genes are also expressed during adulthood, although the distribution of expression is different from that in embryogenesis. For example PAX5 is expressed during haematopoiesis in adult tissues. PAX genes are also expressed in the adult lens tissue of the eye, thymus, thyroid, pancreas, oviduct, vas deferens, epididymis, and in myogenic precursors of muscle tissue.¹

Pax6 is a highly conserved member of a Pax family of genes encoding transcription factors. Mammalian Pax6 genes encode predominantly two forms of the Pax6 protein, Pax6 and Pax6(5a). Pax6 contains a canonical PD. In contrast, Pax6(5a) contains a 14 amino acid insertion within the PAI domain. Pax6 plays critical roles in the organogenesis of the brain, visual, and olfactory systems, as well as peptide hormone gene expression in the pancreas. Pax6 is ubiquitously expressed in the human neural plate and is necessary and sufficient for human neuroectoderm (NE) specification. Pax6 represses pluripotent genes and activates neural genes. Mouse NE specification does not require Pax6.² In addition numerous studies have shown that Pax6 is essential for morphogenesis of the eye from its earliest stages and subsequent formation of all major ocular tissues. PAX6 was shown to act as a migration control gene during development, critical for correct migration of cells into several areas including the eye, olfactory bulb, cortex, and cerebellum. Unlike most other developmental genes, its expression is sustained into adulthood in

certain areas of the brain, including certain septal, diencephalic, and midbrain nuclei, and in astrocytes and certain mature neurons of the dentate gyrus.³ In humans, heterozygous mutations in PAX6 cause a wide spectrum of ocular defects and subtle changes in the olfactory epithelium and brain. In addition, it was found that NeuroD/BETA2 is involved in the activation of the expression of PAX6 through E boxes in the PAX6 promoter.⁴ PAX6 was found to be involved in cancer development. PAX6 was overexpressed in pancreatic carcinoma cells and induced overexpression of Met which promote tumor cell growth. Inhibition of PAX6 transcription lead to a decline in cell growth.⁵ However, PAX6 was shown to suppress the tumorigenicity of glioblastoma multiforme (GBM) cells.

References:

1. Walther, C. et al: Genomics11:424-34, 1991
2. Zhang, X. et al: Cell Stem Cell 7:90-100, 2010
3. Nacher, J. et al: J. Neurosci. Res. 81:753-61, 2005
4. Marsich, E. et al: Biochem. J. 376:707-715, 2003
5. Mascarenhas, J.B. et al: J. Biol. Chem. 284:27524-32, 2009

TECHNICAL INFORMATION

Source:

PAX6 Antibody is a mouse monoclonal antibody raised against recombinant human PAX6 fragments expressed in *E. coli*.

Specificity and Sensitivity:

This antibody detects endogenous PAX6 proteins without cross-reactivity with other family members.

Storage Buffer: PBS and 30% glycerol

Storage:

Store at -20°C for at least one year. Store at 4°C for frequent use. Avoid repeated freeze-thaw cycles.

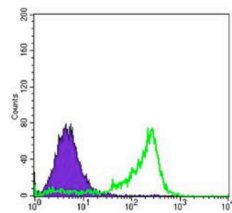
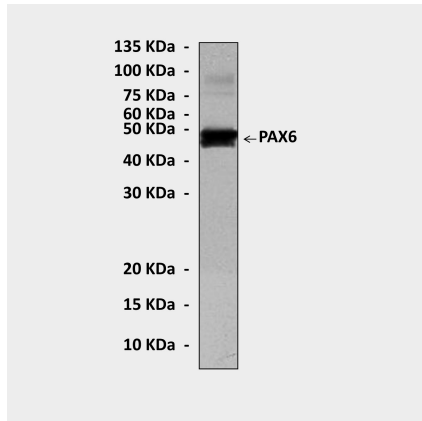
APPLICATIONS

Application:	*Dilution:
WB	1:1000
IP	1:50
IHC	n/d
ICC	n/d
FACS	1:50-200

*Optimal dilutions must be determined by end user.



QUALITY CONTROL DATA



Top: Western Blot detection of PAX6 proteins in HeLa cell lysate using PAX6 antibody. **Bottom:** This antibody also specifically reacts with PAX6 proteins in HeLa cells by FACS testing (PAX6 antibody: Green; control; Purple).

