

Pax-6 (h): 293T Lysate: sc-110018

BACKGROUND

Pax genes contain paired domains with strong homology to genes in *Drosophila* which are involved in programming early development. Lesions in the PAX6 gene accounts for most cases of aniridia, a congenital malformation of the eye, chiefly characterized by iris hypoplasia, which can cause blindness. PAX6 is involved in other anterior segment malformations besides aniridia, such as Peters anomaly, a major error in the embryonic development of the eye with corneal clouding and variable iridolenticulocorneal adhesions. The PAX6 gene encodes a transcriptional regulator that recognizes target genes through its paired-type DNA-binding domain. The paired domain is composed of two distinct DNA-binding subdomains, the amino-terminal subdomain and the carboxy-terminal subdomain, which bind respective consensus DNA sequences. The human PAX6 gene produces two alternatively spliced isoforms that have the distinct structure of the paired domain.

REFERENCES

- Hanson, I.M., et al. 1993. PAX6 mutations in aniridia. *Hum. Mol. Genet.* 2: 915-920.
- Hanson, I.M., et al. 1994. Mutations at the PAX6 locus are found in heterogeneous anterior segment malformations including Peters' anomaly. *Nat. Genet.* 6: 168-173.
- Azuma, N., et al. 1999. Missense mutation in the alternative splice region of the PAX6 gene in eye anomalies. *Am. J. Hum. Genet.* 65: 656-663.
- Fic, W., et al. 2007. Eye development under the control of SRp55/B52-mediated alternative splicing of *eyeless*. *PLoS ONE* 2: E253.
- Yan, Q., et al. 2007. Protein phosphatase-1 modulates the function of Pax-6, a transcription factor controlling brain and eye development. *J. Biol. Chem.* 282: 13954-13965.
- Baer, K., et al. 2007. Sox-2 is expressed by glial and progenitor cells and Pax-6 is expressed by neuroblasts in the human subventricular zone. *Exp. Neurol.* 204: 828-831.
- Xu, H., et al. 2007. Characteristics of progenitor cells derived from adult ciliary body in mouse, rat, and human eyes. *Invest. Ophthalmol. Vis. Sci.* 48: 1674-1682.
- Pinto, G.R., et al. 2007. Mutation analysis of gene PAX6 in human gliomas. *Genet. Mol. Res.* 6: 1019-1025.
- Khan, A.O. and Aldahmesh, M.A. 2008. PAX6 analysis of two unrelated families from the Arabian Peninsula with classic hereditary aniridia. *Ophthalmic Genet.* 29: 145-148.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.

CHROMOSOMAL LOCATION

Genetic locus: PAX6 (human) mapping to 11p13.

PRODUCT

Pax-6 (h): 293T Lysate represents a lysate of human Pax-6 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

Pax-6 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive Pax-6 antibodies. Recommended use: 10-20 µl per lane.

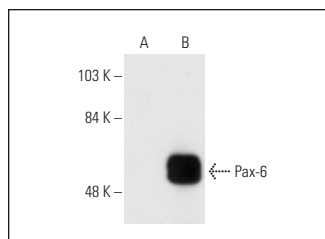
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

Pax-6 (AD2.38): sc-32766 is recommended as a positive control antibody for Western Blot analysis of enhanced human Pax-6 expression in Pax-6 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

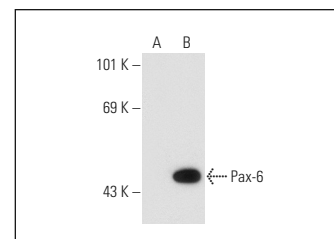
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:
1) Western Blotting: use m-IgGλ BP-HRP: sc-516132 or m-IgGλ BP-HRP (Cruz Marker): sc-516132-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



Pax-6 (AD2.38): sc-32766. Western blot analysis of Pax-6 expression in non-transfected: sc-117752 (A) and human Pax-6 transfected: sc-110018 (B) 293T whole cell lysates.



Pax-6 (AD2.35): sc-53108. Western blot analysis of Pax-6 expression in non-transfected: sc-117752 (A) and human Pax-6 transfected: sc-110018 (B) 293T whole cell lysates.

RESEARCH USE

For research use only, not for use in diagnostic procedures.