

# FOXL2 (A-14): sc-55655

## BACKGROUND

FOXL2 is a 376 amino acid protein encoded by the human gene FOXL2. FOXL2 is found in the nucleus and is believed to be a transcriptional regulator. Defects in FOXL2 are a cause of blepharophimosis, ptosis and epicanthus inversus syndrome (BPES), also known as blepharophimosis syndrome. BPES is an autosomal dominant disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold running inward and upward from the lower lid. In type I BPES (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPES (BPES2) affected individuals show only the eyelid defects. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II. Defects in FOXL2 are also a cause of premature ovarian failure 3 (POF3). POF is a defect of ovarian development and is characterized by hypoenestrogenism, primary or secondary amenorrhea, elevated levels of serum gonadotropins or early menopause. POF is defined as the cessation of ovarian function under the age of 40 years.

## REFERENCES

1. De Baere, E., et al. 2001. Spectrum of FOXL2 gene mutations in blepharophimosis-ptosis-epicanthus inversus (BPES) families demonstrates a genotype-phenotype correlation. *Hum. Mol. Genet.* 10: 1591-1600.
2. Beysen, D., et al. 2004. The human FOXL2 mutation database. *Hum. Mutat.* 24: 189-193.
3. Lee, K., et al. 2005. Transcriptional factor FOXL2 interacts with DP103 and induces apoptosis. *Biochem. Biophys. Res. Commun.* 336: 876-881.
4. Raile, K., et al. 2005. A new heterozygous mutation of the FOXL2 gene is associated with a large ovarian cyst and ovarian dysfunction in an adolescent girl with blepharophimosis/ptosis/epicanthus inversus syndrome. *Eur. J. Endocrinol.* 153: 353-358.
5. Vincent, A.L., et al. 2005. Blepharophimosis and bilateral Duane syndrome associated with a FOXL2 mutation. *Clin. Genet.* 68: 520-523.
6. Or, S.F., et al. 2006. Three novel FOXL2 gene mutations in Chinese patients with blepharophimosis-ptosis-epicanthus inversus syndrome. *Chin. Med. J.* 119: 49-52.
7. Nakamoto, M., et al. 2006. Molecular cloning and analysis of gonadal expression of FOXL2 in the medaka, *Oryzias latipes*. *Biochem. Biophys. Res. Commun.* 344: 353-361.
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## CHROMOSOMAL LOCATION

Genetic locus: FOXL2 (human) mapping to 3q22.3; Foxl2 (mouse) mapping to 9 E3.3.

## SOURCE

FOXL2 (A-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of FOXL2 of mouse origin.

## PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-55655 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

FOXL2 (A-14) is recommended for detection of FOXL2 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

FOXL2 (A-14) is also recommended for detection of FOXL2 in additional species, including canine.

Suitable for use as control antibody for FOXL2 siRNA (h): sc-106837, FOXL2 siRNA (m): sc-77399, FOXL2 shRNA Plasmid (h): sc-106837-SH, FOXL2 shRNA Plasmid (m): sc-77399-SH, FOXL2 shRNA (h) Lentiviral Particles: sc-106837-V and FOXL2 shRNA (m) Lentiviral Particles: sc-77399-V.

Molecular Weight of FOXL2: 38 kDa.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

## SELECT PRODUCT CITATIONS

1. Farhat, A., et al. 2011. Hematopoietic-Prostaglandin D2 synthase through PGD2 production is involved in the adult ovarian physiology. *J. Ovarian Res.* 4: 3.

## STORAGE

Store at 4° C, \*\*DO NOT FREEZE\*\*. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## RESEARCH USE

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



Try **FOXL2 (262C1a): sc-81275**, our highly recommended monoclonal alternative to FOXL2 (A-14).