

# FOXL2 (262C1a): sc-81275

## 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 hypogonadism, 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

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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.

## SOURCE

FOXL2 (262C1a) is a mouse monoclonal antibody raised against a recombinant protein corresponding to an internal region of FOXL2 of human origin.

## PRODUCT

Each vial contains 100 µg IgG<sub>1</sub> kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 1.0% stabilizer protein.

## APPLICATIONS

FOXL2 (262C1a) is recommended for detection of FOXL2 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) and immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)].

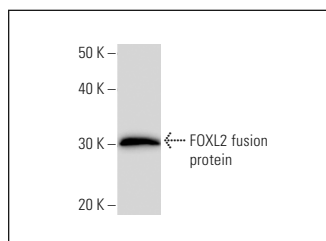
Suitable for use as control antibody for FOXL2 siRNA (h): sc-106837, FOXL2 shRNA Plasmid (h): sc-106837-SH and FOXL2 shRNA (h) Lentiviral Particles: sc-106837-V.

Molecular Weight of FOXL2: 38 kDa.

## RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-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. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).

## DATA



FOXL2 (262C1a): sc-81275. Western Blot analysis of human recombinant FOXL2 fusion protein.

## STORAGE

For immediate and continuous use, store at 4° C for up to one month. For sporadic use, freeze in working aliquots in order to avoid repeated freeze/thaw cycles. If turbidity is evident upon prolonged storage, clarify solution by centrifugation.

## RESEARCH USE

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

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.