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 hypoestrogenism, 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


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.