CLINICAL CASE DESCRIPTION

Patient: girl, 14 years 6 months old, complained of irregular menstrual cycle.

Clinical examination: sexual development Tanner 4, Me at 13 years old, 3 menses, with a last delay up to 8 months. Single terminal hairs along the white line of the abdomen, areola. The patient had epicanthus and narrowed eye slits of different sizes. She was operated on for congenital blepharophimosis in infancy. No one has similar problems in the family.

The results of hormonal examination are presented in the table.

Pelvic organs ultrasound: endometrial thickness is 0.8 cm, yellow body cyst of the right ovary were revealed (echographic signs of a fluid formation with a diameter of 3.0 cm in the right ovary).

During the inpatient examination the girl underwent the trial therapy with dydrogesterone (Duphaston). According to hormonal studies, provided on the 4th day of the menstrual cycle, hypergonadotropic hypogonadism is revealed (the results are in the table).

Pelvic organs ultrasound showed normalization of the ovarian volume, follicle persistence in the right ovary (left ovary volume 4.51 ml, right ovary volume 5.84 ml, endometrial thickness 0.45 cm).

The patient was recommended with a combined oral contraceptive therapy (ethinyl estradiol + desogestrel), which the girl's mother refused.

Medical examination after 3 months without treatment revealed hypergonadotropic hypogonadism (the results of hormonal examination are presented in the table). The measured level of antimullerian hormone was low.

Considering the combination of blepharophimosis, hypergonadotropic hypogonadism, a defect in the FOXL2 gene was suspected. The girl underwent the molecular-genetic research.