P2-P50 Ovarian cysts in a 46,XX patient with congenital lipoid adrenal hyperplasia and with spontaneous puberty

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BACKGROUND

Congenital lipoid adrenal hyperplasia (CLAH) is the most severe form of congenital adrenal hyperplasia, characterized by lack of synthesis of all kinds of steroids in adrenals and gonads due to defects in gene of Steroidogenic Acute Regulatory protein (StAR). 46,XX patients can have a spontaneous puberty due to residual estrogen synthesis by a StAR-independent pathway in ovary. Development of ovarian cysts may be derived from persistent anovulation and impairment of ovarian StAR-independent steroidogenesis by lipoid deposition.

CASE REPORT

We report a 13-years-old 46,XX patient with CLAH, which presented a spontaneous puberty and enlarged policystic ovaries. A congenital adrenal insufficiency was diagnosed due to hyponatremia, hyperkalemia and hyperpigmentation at 3 weeks after birth. She was successfully treated with hydrocortisone and fludrocortisone. The decreased level of all steroid hormones was determined by multisteroid analysis. Two heterozygous mutations (466delG and W245X) were detected in the StAR gene. Brest development was observed at the age of 11. It progressed to the Tanner stage 4 by 13,5 years old, but no pubic and axillary hair was observed. At the age of 13 giant cyst in the right ovary (200 cm3) was revealed by ultrasound, which was successfully treated with cyclic progesterone therapy. Estrogen therapy was not prescribed because of normal gonadotropins and estradiol levels (Tab.1). After 5-month of treatment with progesterone the increase of LH level (20.9 U/l) was detected and enlarged polycystic ovaries was revealed by MRI (right ovary 38×35×38 mm; left ovary 35×28×36 mm). The combined therapy with estrogen and progesterone was started.

Table 1. Clinical and laboratory data of female patient with congenital lipoid adrenal hyperplasia (CLAH)

Age	Tanner stage	LH, U/l	FSH, U/l	Estradiol, pmol/l	Ovaries, cm3	Progesterone or estrogen treatment
11. y	Brest 2 Pubis 1	**0,2** (1,5-4,1)	**5,3** (1,0-10,8)	**45** (35-90)	Ovarian cyst 1,8 cm	
12 y	Brest 3 Pubis 1	**0,6** (2,0-6,3)	**3,2** (1,5-12,8)	**40** (50-220)	Ovaries volume 4,5 cm3	
13 y	Brest 3 Pubis 1	**5,89** (2,0-6,3)	**4,16** (1,5-12,8)	**221,9** (50-220)	Ovarian cyst **200** cm3	Cyclic Progesterone therapy
13,5 y	Brest 4 Pubis 1	**20,9** (3,2-9,8)	**10,1** (1,5-11,7)	**101,7** (80-330)	Polycistic ovaries **21,6** cm3 and **26,2** cm3	Combined therapy with Estrogen+Progesterone

CONCLUSIONS

Ovarian cysts can be seen in 46,XX CLAH patients at puberty. Replacement therapy with estrogen and progesterone can prevent the ovarian cysts formation. The long-term follow-up is necessary in female patients with CLAH.

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