KLINEFELTER SYNDROME WITH LOW GONADOTROPIN LEVELS

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BACKGROUND

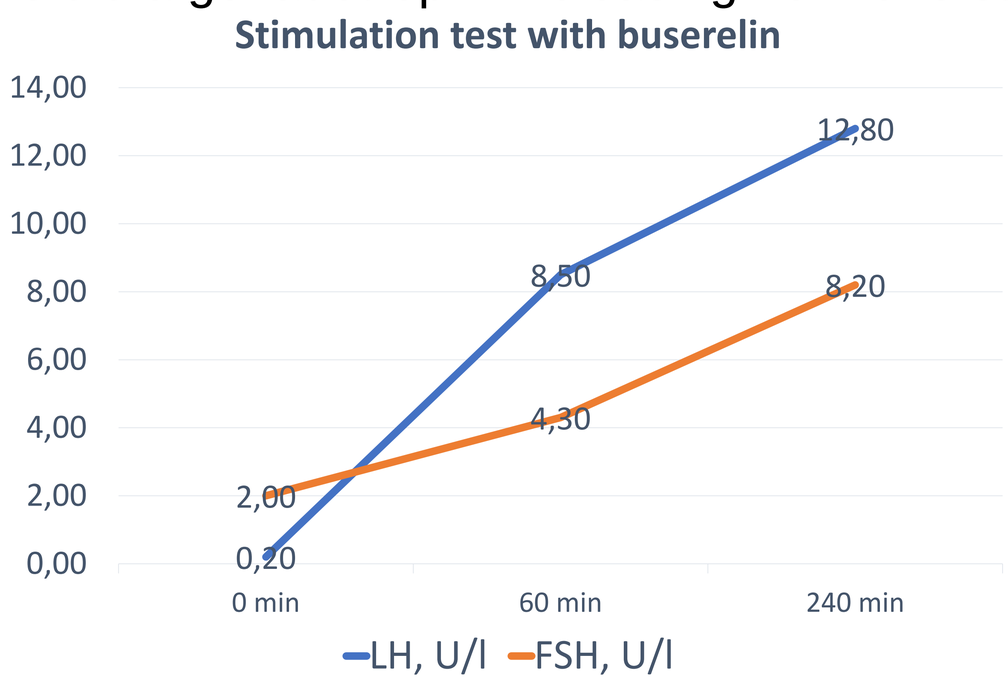
Klinefelter syndrome (KS) is the most common cause for hypergonadotropic hypogonadism. Patients with 47,XXY karyotype often have increased gonadotropin levels at early puberty, which stay high during adolescent and adult life due to hyalinisation of seminiferous tubules of testes. We report a clinical case of 47,XXY KS patient with hypogonadism and low gonadotropin levels.

CLINICAL CASE

A boy was referred to an endocrinologist at the age of 12,5 years due to overweight and small testis. Physical examination revealed: height 158.4 cm (SDS=0.58), BMI=23.23 kg/м2 (SDS=+1.38), Tanner stage 1 with no signs of eunuchoid body shape. The testis were soft and the testicular volume was 1 ml. Bone age was 13 years (G&P).
We tested his blood for hormone levels:

LH, U/l	FSH, U/l	Basal testosterone, nmol/l	Testosterone after HGH stimulation test, nmol/l	Cortisol at 8 a.m., nmol/l	Free T4, pmol/l	TSH, mlU/l
0.2	1.8	0.6	4.5	415.3	15.3	2.74

Levels of LH, FSH were estimated as normal, while basal and stimulated testosterone levels – as low. The patient was found to have normal levels of 8:00 cortisol, free T4, thyroid-stimulating hormone, there were no complaints of headache, no signs of space-occupying lesion in his brain.
We did a gonadotrophin releasing hormone stimulation test with buserelin:



The test didn't reveal the hypogonadotropic hypogonadism (LH peak 12,8 U/l, FSH peak 8,2 U/l).
The diagnosis of KS was confirmed by karyotype (47,XXY, [100]) and fluorescence in situ hybridization method (47,XXY nuc ish (DXZ1x2,DYZ3x1, SRYx1)[100]).

Testosterone therapy was initiated with the starting dose of 50 mg 1 time per 4 weeks with further increase to 250 mg 1 time per 4 weeks. After a year of treatment levels of LH and FSH stayed low:

LH, U/l	FSH, U/l	Basal testosterone, nmol/l	Inhibin B, pg/ml	AMH, ng/ml
0.9	3.6	9.0	51.1	75.9

Conclusions

There are very few cases of KS without hypergonadotropic hypogonadism described today. Low gonadotropin levels could be explained by significant variability of manifestations of KS even with classical karyotype.

Conflict of interests

Authors declair no conflicts of interests.