OBJECTIVE

To evaluate clinical and laboratory characteristics of various types of gonadal dysgenesis in girls with hypergonadotrophic hypogonadism.

METHODS

17 girls (13.9 ± 3.72 years) with hypergonadotrophic hypogonadism were examined. Inclusion criteria: characteristics of delayed puberty, female phenotype, presence of müllerian duct derivatives, high levels of gonadotrophins. Tanner stage, antropometric data, bone ages, genitometric parameters, LH, FSH, estradiol (E2), testosterone (T) levels, cytogenetic and molecular genetic tests were provided. Results were evaluated according to the reference rages in girls.

RESULTS

The patients were divided into 3 groups:

- 45,X (8/17)
- 46, XY (5/17)
- 46,XX (4/17)

There were no differences between groups in uterus volumes (Me 4.24 vs 1.8 vs 2.1 ml, p>0.05) and in gonadotrophins levels (Me LH 25.1 vs 12.57 vs 25.3 uUI/ml, p>0.05; Me FSH 56.01 vs 88.4 vs 108.7 uUI/ml, p>0.05).

Girls with Y chromosome had upper normal height (Me SDS 1.66), 46,XX had average height (Me SDS 0.02, p=0.027) and girls 45,X had low height (Me -3.46, p=0.003, growth failure rate was 87.5% (in 7/8 girls, p=0.01).

There was no difference between bone ages in girls 46,XY (Me SDS -1.6) and 46,XX (Me SDS -1.9, p=0.325), which were low normal while there was delayed bone ages in girls 45,X (Me SDS bone age – 3.74, p=0.027).

Serum E2 levels in girls 46,XY were higher (Me 44.81) compared with girls 46,XX (Me 13.75, p=0.013) and with girls 45,X (Me 11.29, p=0.028), while there was no difference between two last groups (p=0.82).

Serum T levels in girls with Y chromosome were elevated (Me T 4.6) compared with the levels of same-aged and were higher compared with girls 46,XX (Me T 0.25, p=0.015).

Molecular genetic test were provided in 46,XY. Heterozygote mutation of gene WT1 was diagnosed in 1 girl.

CONCLUSION

The following types of gonadal dysgenesis in girls with hypergonadotrophic hypogonadism were diagnosed: Turner syndrome, pure gonadal dysgenesis (46 XY, 46 XX). Among 3 groups there were significant differences in girls with Y chromosome: upper normal values of height, more progressed stage of puberty and elevated estrogen and testosterone concentrations. These features can be caused by germ cell tumor.