CLINICAL AND LABORATORY CHARACTERISTICS OF PATIENTS WITH DIFFERENT VARIANTS OF GONADAL DYSGENESIS

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OBJECTIVE

To study clinical and laboratory characteristics of patients with disorders of sex development (DSD) 45,X/46,XY and 46,XY, partial gonadal dysgenesis (PGD)

SUBJECTS and METHODS

It was included 27 patients with disorders of gonadal dysgenesis since birth before 9 years. Gonadal dysgenesis criteria:
- 45,X/46,XY
- 46,XY with derivates Mullerian duct
All children evaluated:
- The structure of external genitalia by the external masculinization score (EMS, range 0-12)
- The structure of the internal genitalia (pelvic ultrasound, n=27, laparoscopy, n=25)
- Hormonal research in mini-puberty (follicle-stimulating hormone, FSH, n=15, luteinising hormone, LH, n=14; inhibin B, n=9)
- Hormonal research in mini-puberty and neutral period (anti-Mullerian hormone, AMH, n=24, basal testosterone and after the human chorionic gonadotrophin stimulation test, ΔT; n=22)

RESULTS

1. All patients were divided into two groups based on cytogenetic survey
- 45,X/46,XY
- 46,XY

2. Age verification diagnosis in patients were divided into three groups
- up to 1 month
- up to 1 year
- up to 3 years

3. Male gender selected in 76% of patients with mosaicism and in 60% with partial gonadal dysgenesis

CONCLUSION

Patients with DSD 45,X/46,XY in comparison with DSD 46,XY partial gonadal dysgenesis had safer gonad function and more pronounced degree of masculinization of the external genitalia