



# 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 y.o

Gonadal dysgenesis criteria:

- 45,X/46,XY
- 46,XY with derivats 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

All patients were divided into two groups based on cytogenetic survey

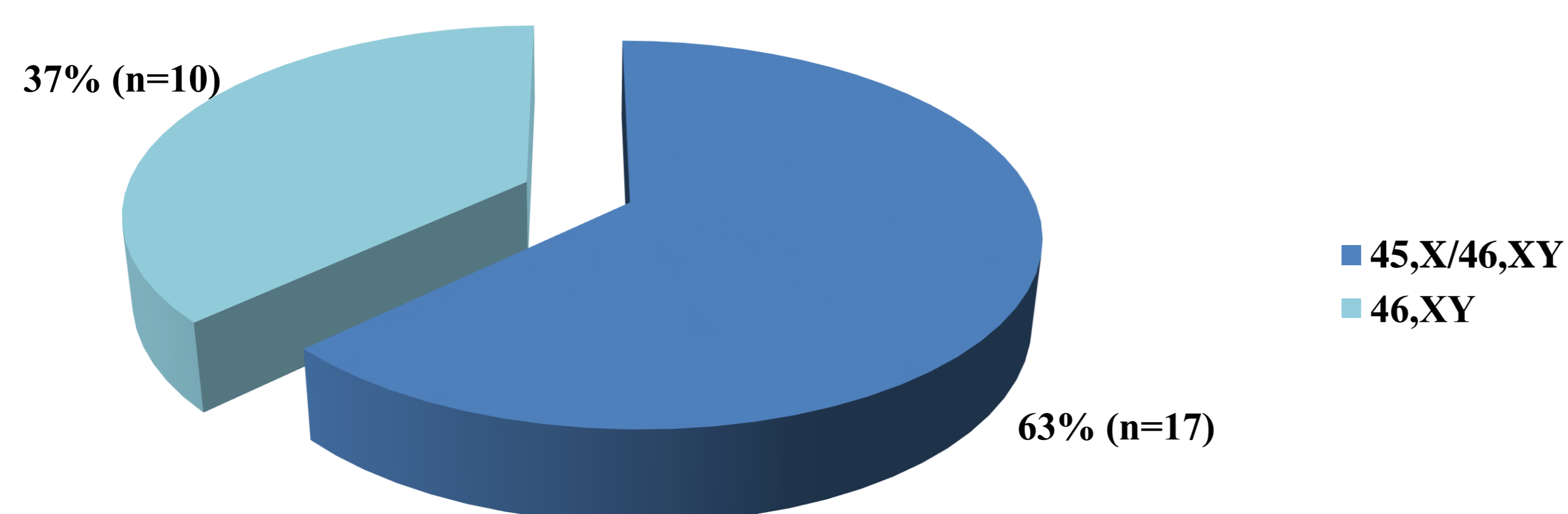


Figure 1. The structure of patients with DSD

Age verification diagnosis in patients were divided into three groups

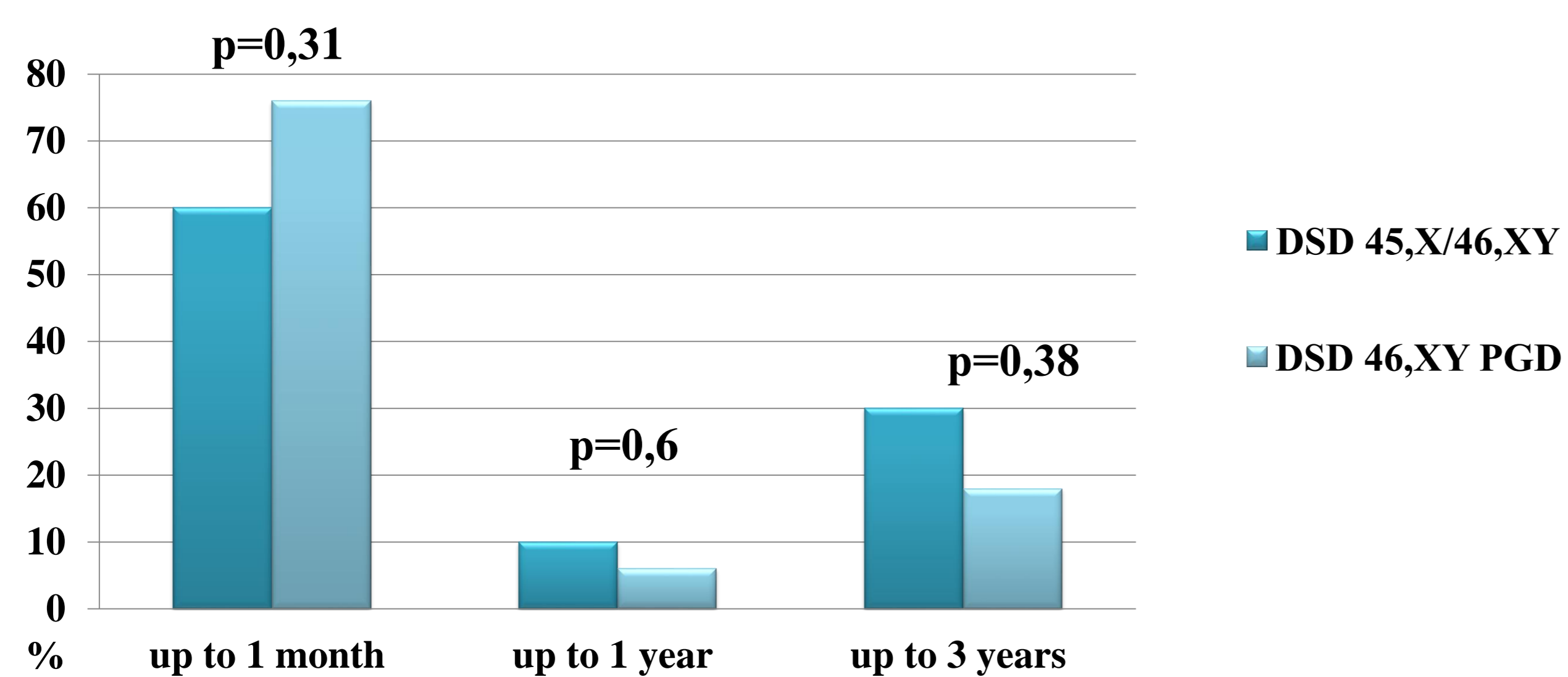


Figure 2. Age verification diagnosis

Male gender selected in 76% of patients in group with mosaicism and in 60% - with partial gonadal dysgenesis

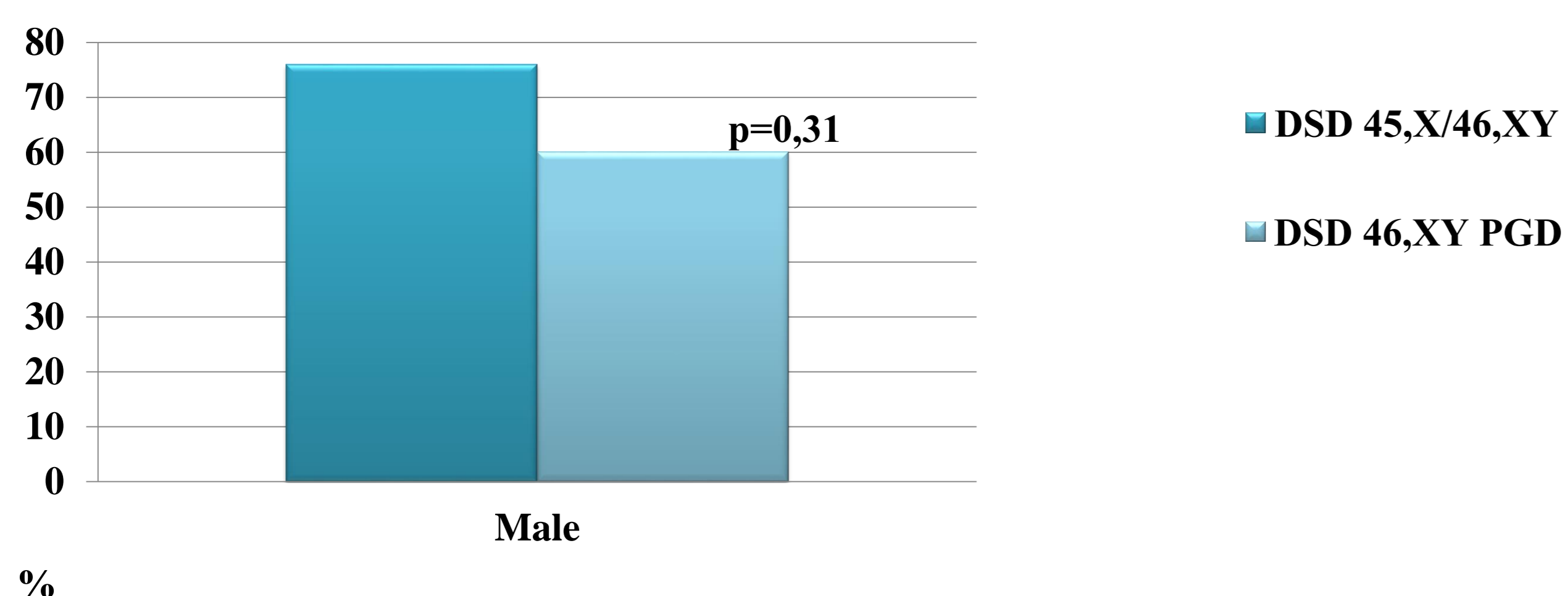


Figure 3. The male sex of rearing in patients

The reason for the initial treatment of all patients was ambiguous genitalia

Mediana EMS:

4,5 [1;10] in patients with mosaicism

1,25 [1;5] – with DSD 46,XY, partial gonadal dysgenesis (p=0,033)

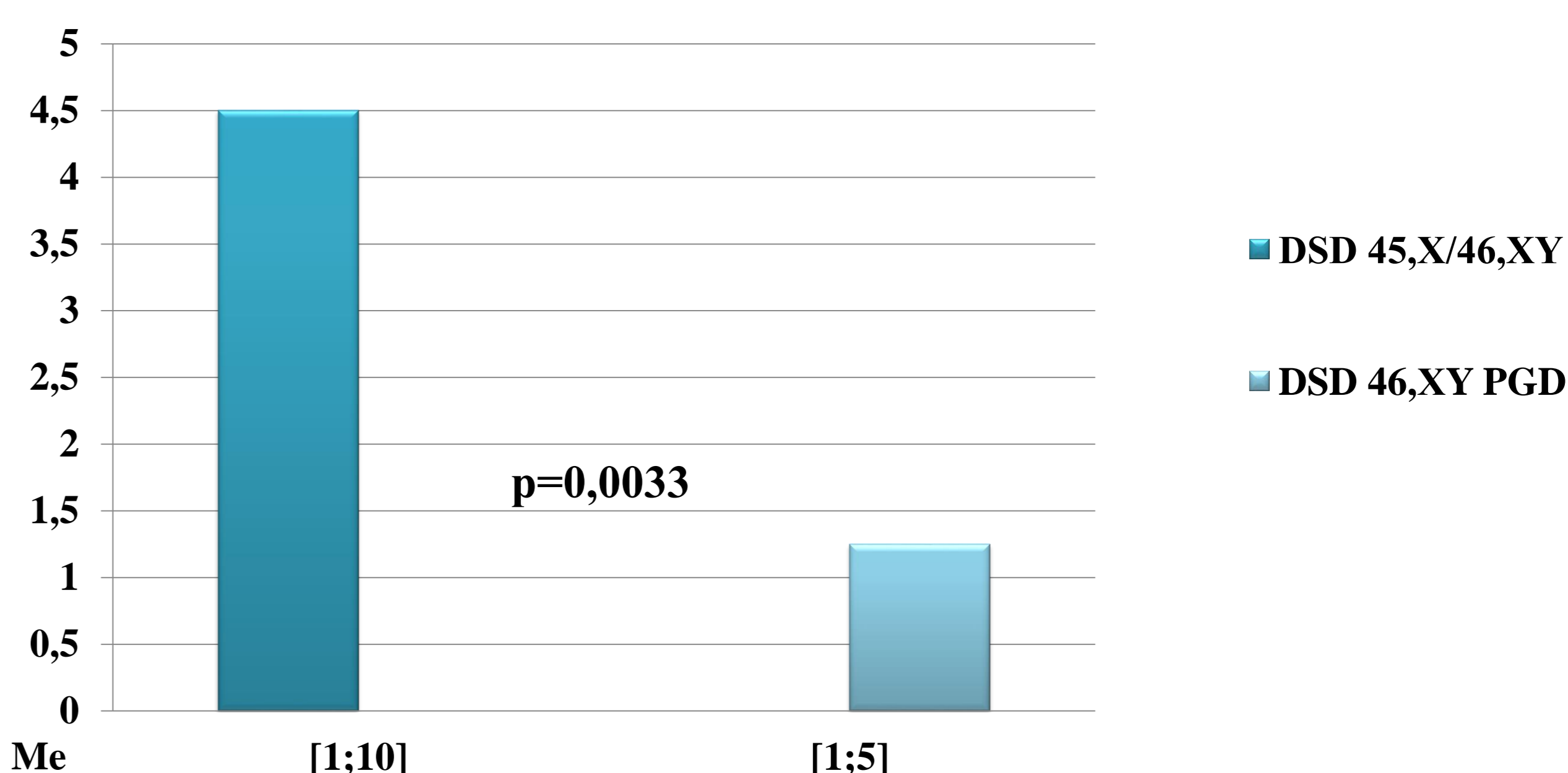


Figure 4. The structure of the external genitalia on the external masculinization score

In the group with DSD 46,XY PGD were more often elevated values FSH and LH than in the group with DSD 45,X/46,XY

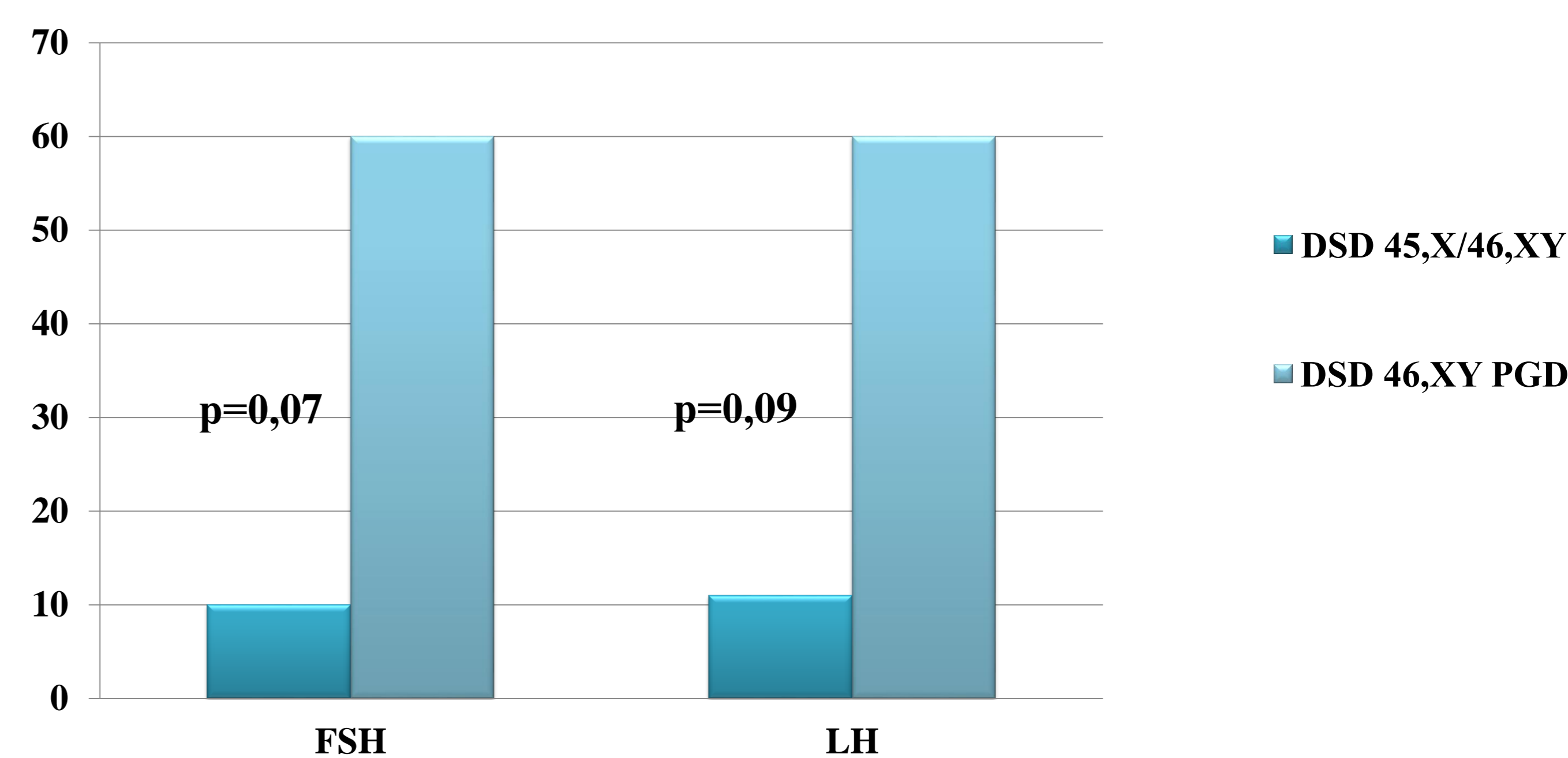


Figure 5. Functional state of the pituitary-gonadal system

Functional state of the gonads in patients with mosaicism was more safe than in patients with 46,XY PGD

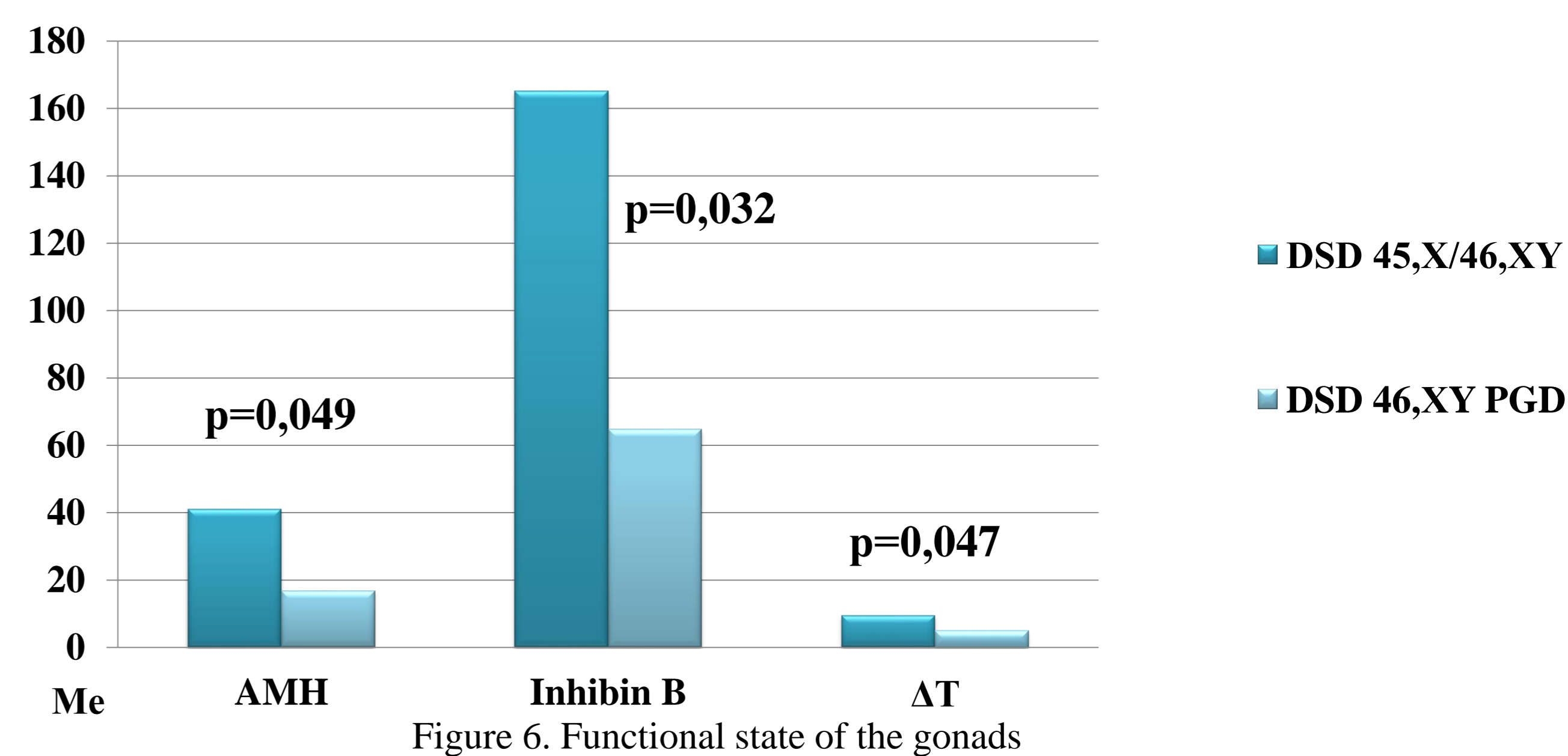


Figure 6. Functional state of the gonads

## 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

