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OBJECTIVE

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

SUBJECTS and METHODS

It was included 248 patients with genital ambiguity since birth before 18 years old.

All children with mosaicism 45,X/46,XY evaluated

- The structure of the external genitalia on the external masculinization score (EMS, range 0-12)
- Ultrasound examination
- The definition of anti-Mullerian hormone (AMH, n=15)
- Basal and stimulated human chorionic gonadotropin testosterone (T, n=12)

We removed 7 gonads of 11 patients

RESULTS

All patients were divided into groups based on cytogenetic survey:

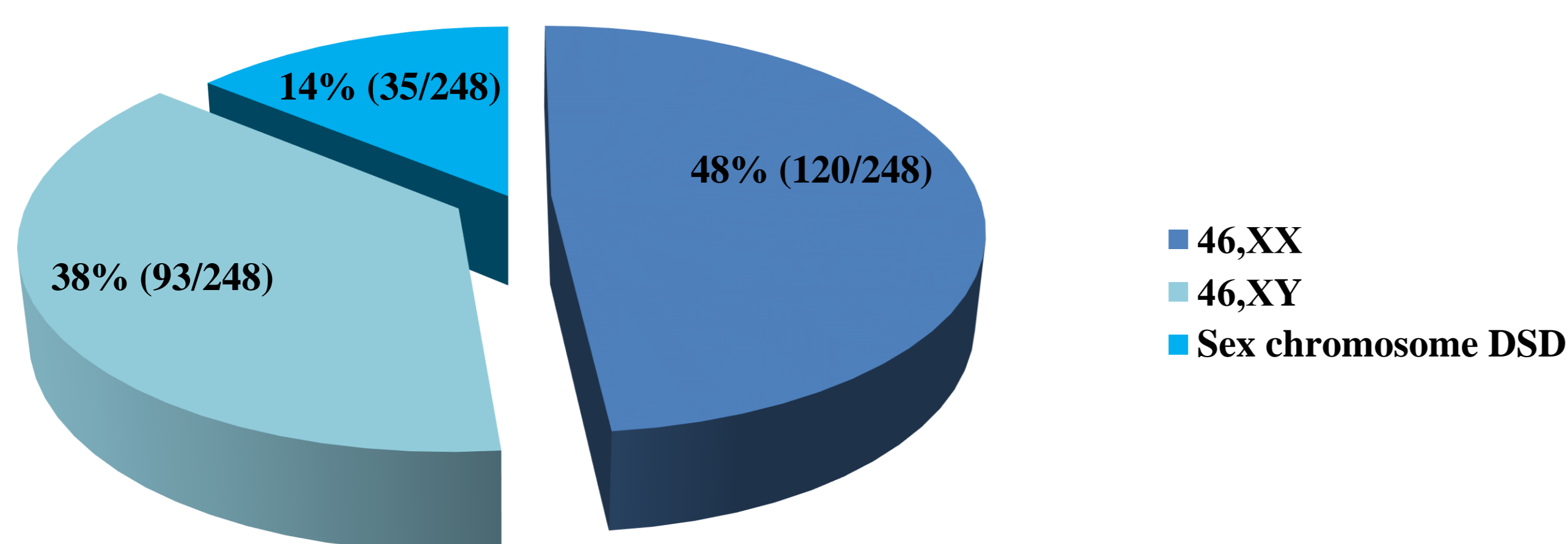


Figure 1. The structure of patients with DSD

Patients with sex chromosome DSD had next variants:

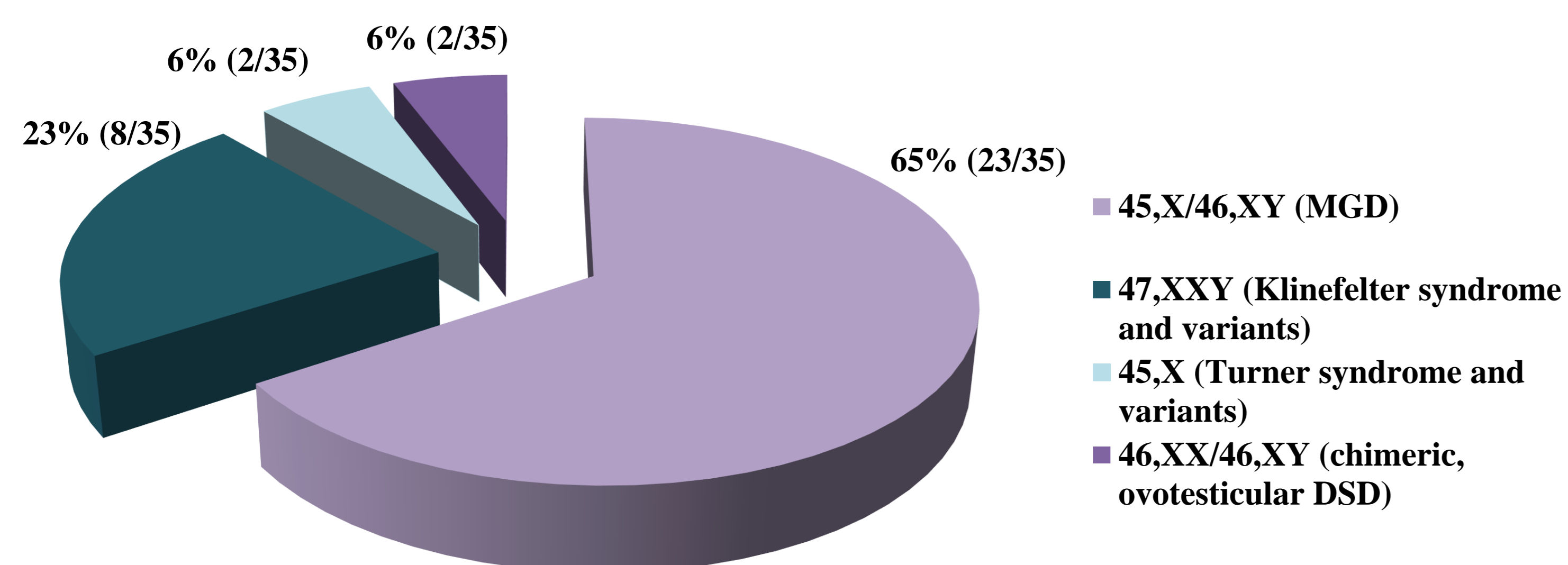


Figure 2. The structure of patients with sex chromosome DSD

The sex of rearing patients with mosaicism 45,X/46,XY

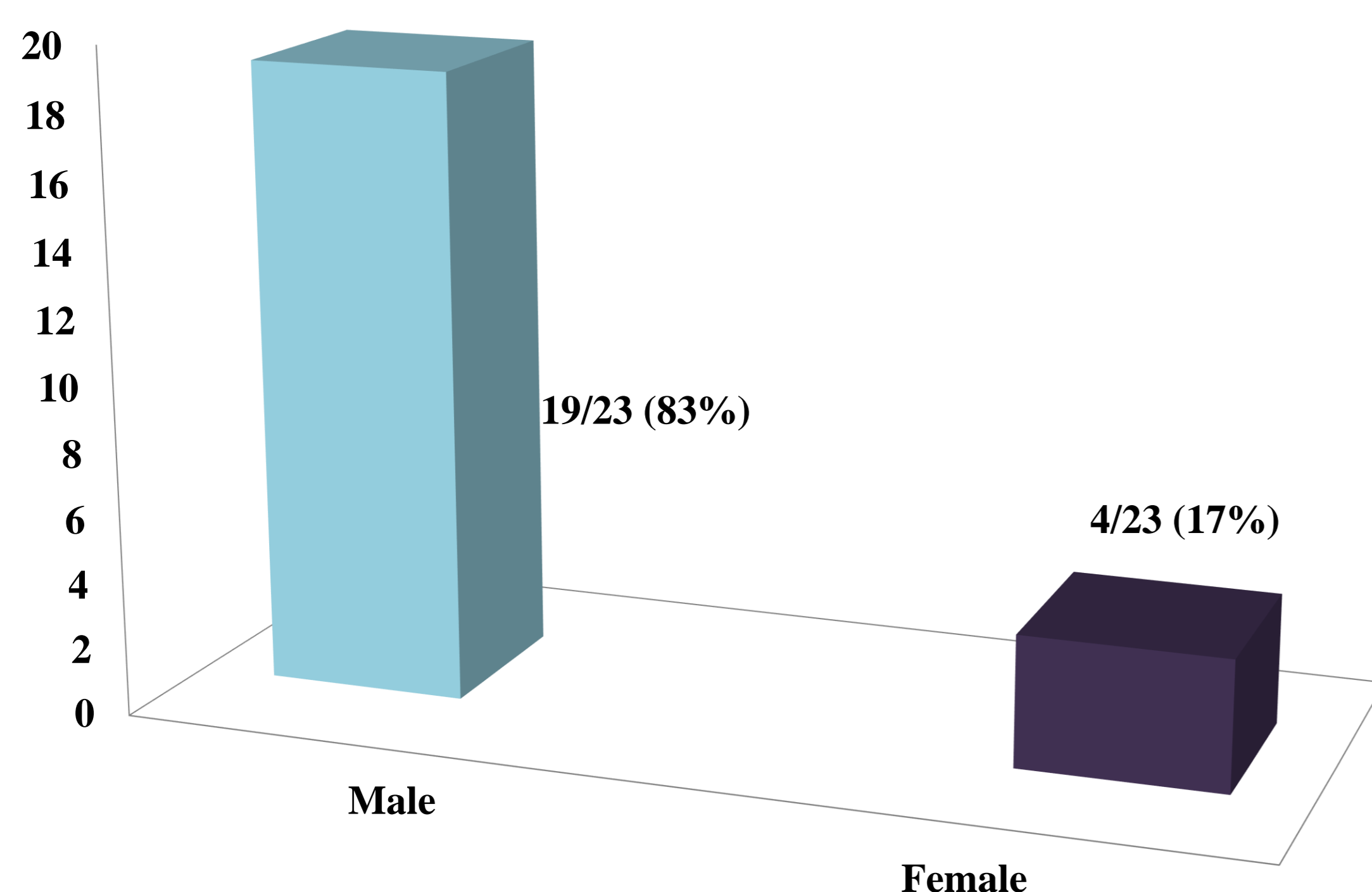


Figure 3. The sex of rearing patients with mosaicism

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

Mediana EMS was 3 [1÷11].

Range of EMS of 17 male patients was from 1 to 11, all female patients was 1.



Picture 1. Ambiguous genitalia

Mullerian remnants were revealed in 86% (18/21).

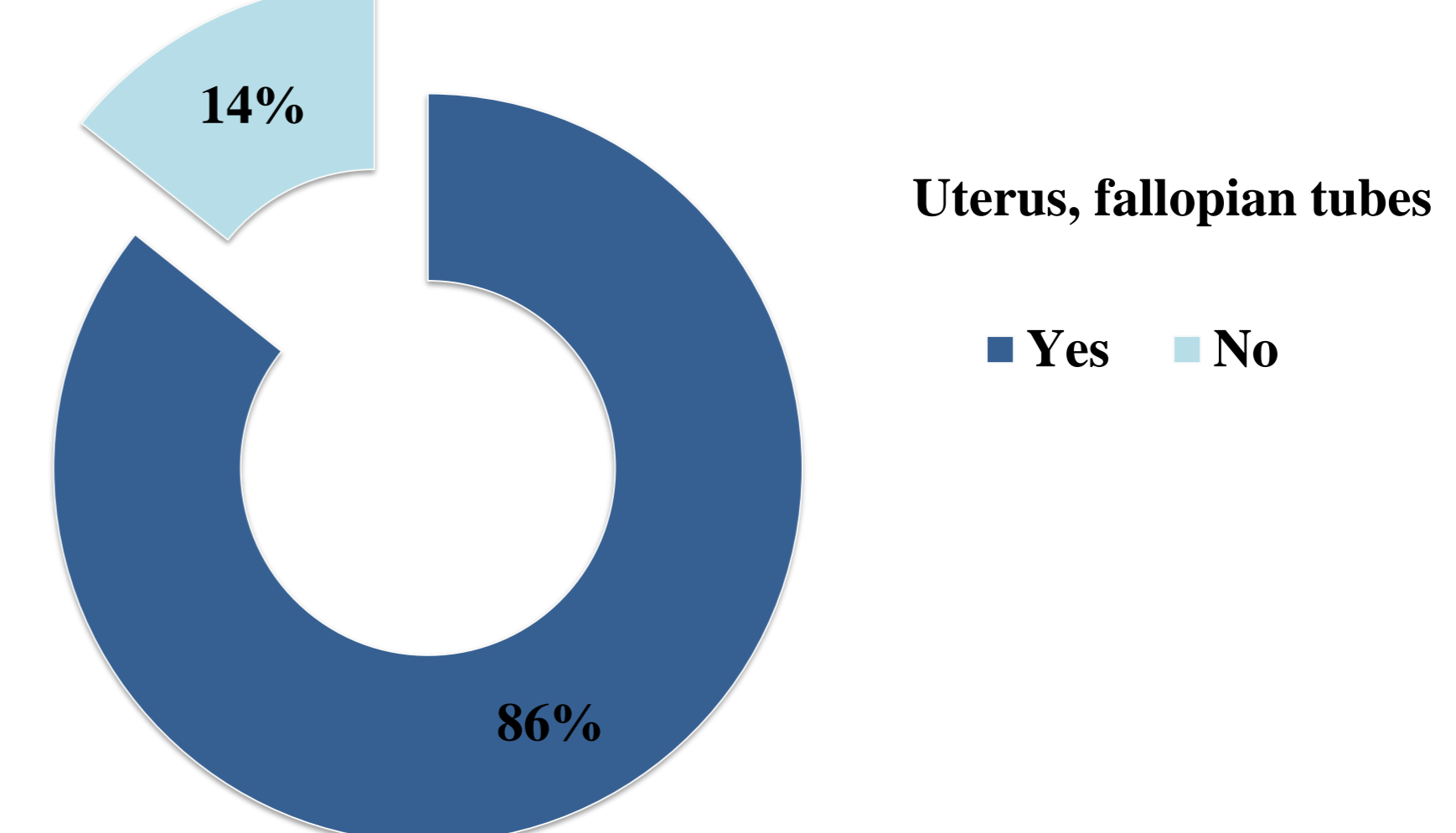


Figure 4. Mullerian remnants by ultrasound examination

Gonadal examination of 7 gonads showed classical picture of mixed gonadal dysgenesis had just 28,5% (2/7) of cases.

Patients with 45,X/46,XY frequently show stigmata typically associated with Turner syndrome

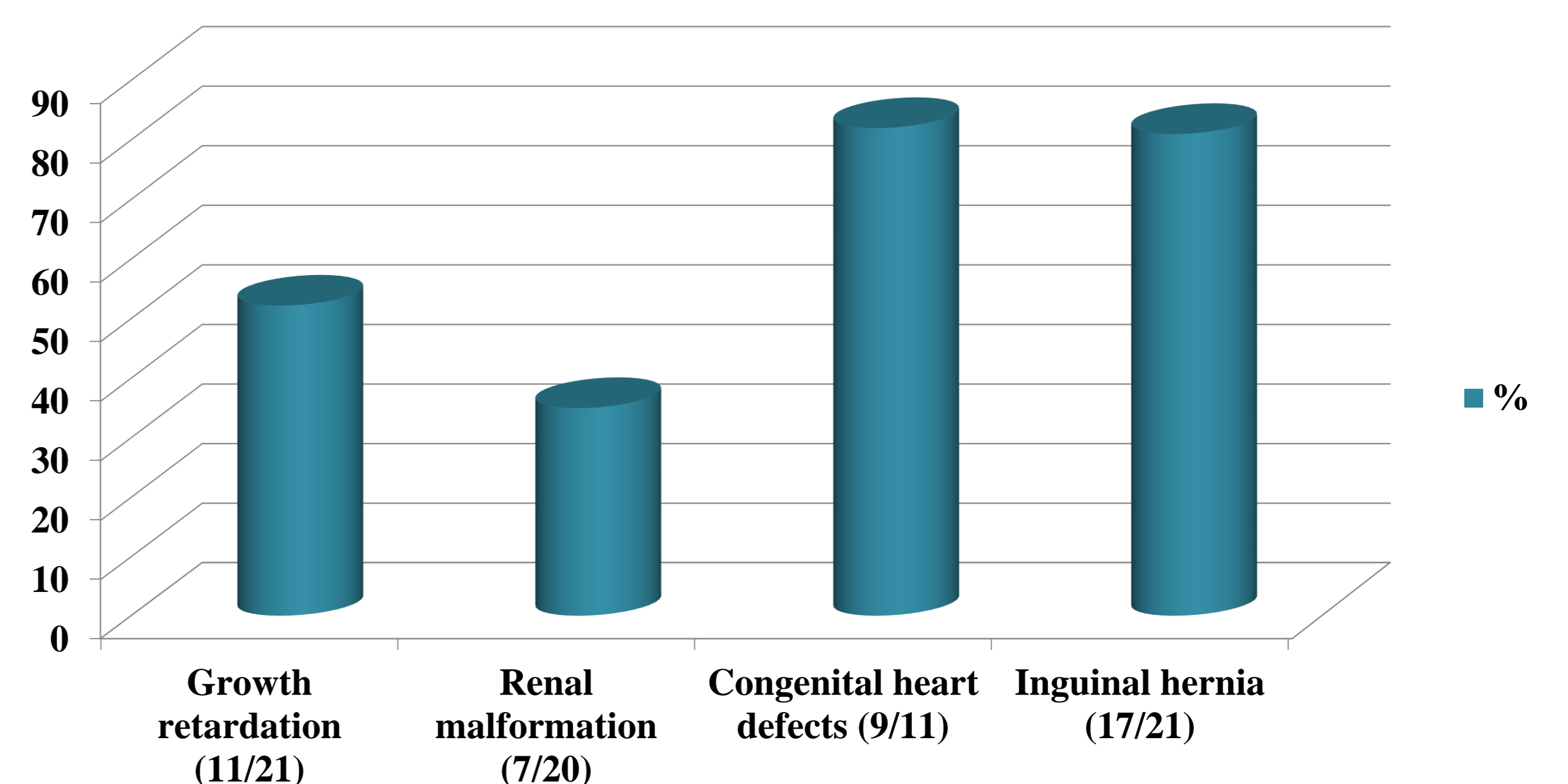


Figure 5. Congenital malformation from patients with DSD 45,X/46,XY

During hormonal evaluation was detected positive correlation between basal Testosterone in mini-puberty and range EMS (n=8, p=0,01).

There was a trend to higher frequency low anti-Mullerian hormone compared to the frequency poor Testosterone response to the test with human chorionic gonadotropin (n=12, p=0,17).

CONCLUSION

- ❖ The group of patients with DSD 45,X/46,XY was heterogenous in structure of external genitalia, internal genitalia and degree of gonadal dysgenesis.
- ❖ In most cases patients had low levels of anti-Mullerian hormone, which is a more significant marker of testicular dysgenesis than ΔT .
- ❖ We detected positive correlation between basal Testosterone in mini-puberty and range EMS.