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:
  - 46,XX
  - 46,XY
  - Sex chromosome DSD
  - 45,X/46,XY (MGD)
  - 47,XXY (Klinefelter syndrome and variants)
  - 45,X (Turner syndrome and variants)
  - 46,XX/46,XY (chimeric, ovotesticular DSD)
- Patients with sex chromosome DSD had next variants:
  - 45,X/46,XY (MGD)
  - 47,XXY (Klinefelter syndrome and variants)
  - 45,X (Turner syndrome and variants)
  - 46,XX/46,XY (chimeric, ovotesticular DSD)
- The sex of rearing patients with mosaicism 45,X/46,XY

CONCLUSION
- 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.
- Mullerian remnants were revealed in 86% (18/21).
- 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
- During hormonal evaluation was detected positive correlation between basal Testosterone in mini-puberty and range EMS.
  - 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=0,01).
- We detected positive correlation between basal Testosterone in mini-puberty and range EMS.