# ETIOLOGICAL STRUCTURE DISORDERS OF SEX DEVELOPMENT 46,XY BY ONE CENTER

O.Y.Latyshev[1], E.S.Sannikova[1], L.N.Samsonova[1], E.V.Kiseleva[1], G.F.Okminyan[1], E.P.Kasatkina[1], O.M.Dondup[2], A.B.Okulov[1], A.N.Tulpakov[3]

[1]Russian Medical Academy of Postgraduated Education Study, Moscow, Russia

[2] Pirogov Medical University, Moscow, Russia

[3]National Medical endocrinology research center, Moscow, Russia

## OBJECTIVE

**To study structure disorders of sex development (DSD) 46,XY by one center**

## SUBJECTS and METHODS

**It was included 60 patients with diagnosis DSD 46,XY at birth to 18 years.**

**For all patients was conducted structural evaluation of the external (by External Masculinization Score, EMS, 0-12) and internal genitalia (by pelvic ultrasound, n=60, laparoscopy, n=20),**
**hormonal research (testosterone, dihydrotestosterone, androstendione, anti-Mullerian hormone, AMH, inhibin B, follicle-stimulating hormone, luteinising hormone) in mini-puberty (n=28), neutral period (n=21) and puberty (n=11), molecular genetic studies Ion Torrent custom Ampliseq_DSD (n=37) and gene such us AR (n=14), SF1 (n=2), SRY (n=3), CYP21 (n=2), WT1 (n=2), histology of gonads removed (n=23 by 15 patients).**
**Gonadal dysgenesis criteria: derivats Mullerian duct, AMH < 55 ng/ml in mini-puberty and AMH < 85 ng/ml in neutral period.**

## RESULTS

- **A definitive diagnosis was received in 56% (33/60) of children with 46,XY DSD**

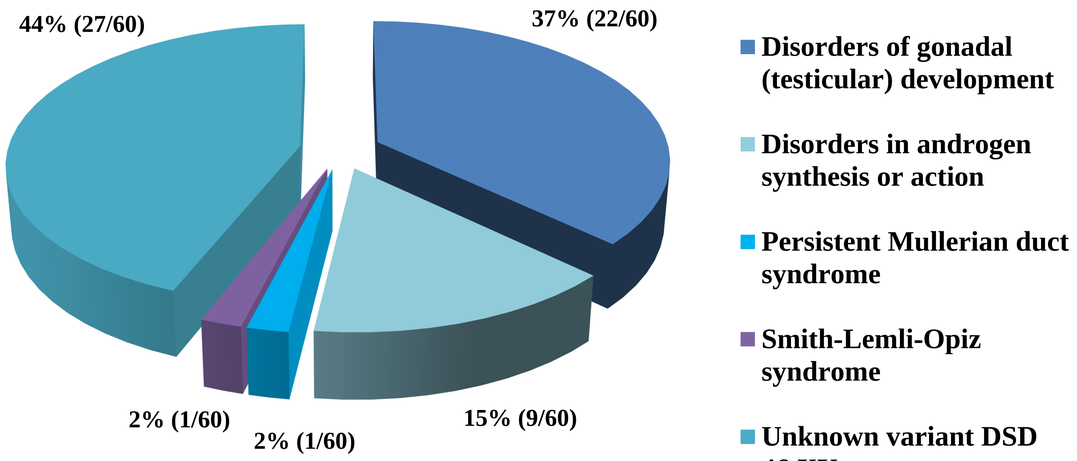


Figure 1. The structure of patients with DSD 46,XY

- **Disorders of gonadal development include complete gonadal dysgenesis , partial gonadal dysgenesis and ovotesticular DSD**

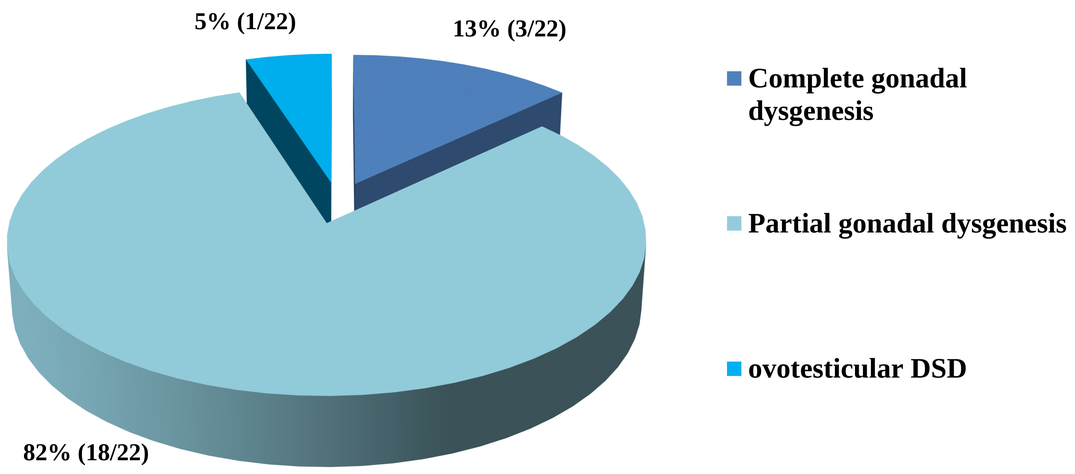


Figure 2. The structure of patients with DSD 46,XY gonadal development

- **Disorders in androgen synthesis or action presented by total and partial androgen insensitivity syndrome**

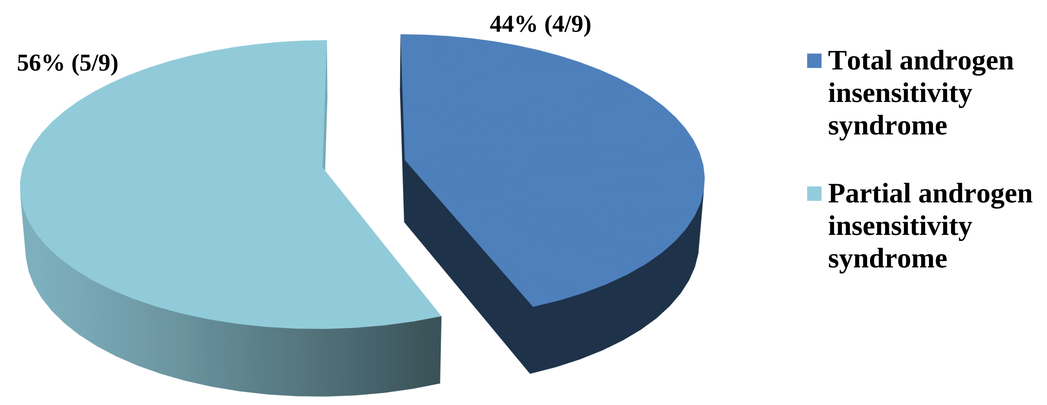


Figure 3. The structure of patient with Disorders in androgen synthesis or action presented by total and partial androgen insensitivity syndrome

- **Mutations in genes in patients with nosological variant DSD 46,XY**

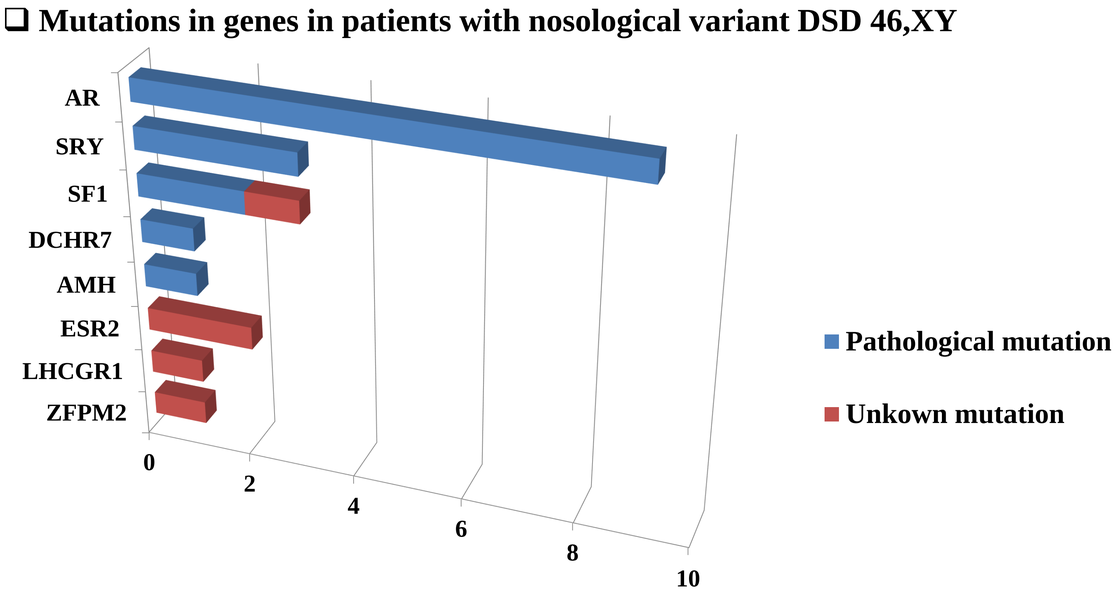


Figure 4. The structure of genes with mutations in patients with DSD 46,XY

- **While y 44% (27/60) of patients didn't have verified variant of nosological pathology and 5 patients had mutation in genes with pathological significance of today is not known**

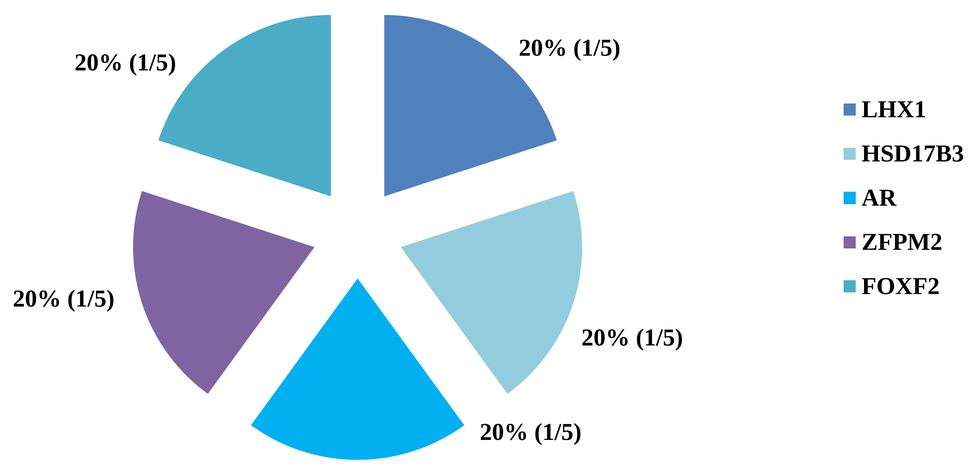


Figure 4. The structure of genes with mutations in patients with DSD 46,XY without nosological variant

## CONCLUSION

**Completed complex survey including molecular genetic analysis allowed to verify nosological variant of DSD 46,XY only in 56% (33/60) of patients.**
**Rating of nosological variants of DSD 46,XY by frequency: partial gonadal dysgenesis (67%, 22/33), androgen insensitivity syndrome (27%, 9/33), total gonadal dysgenesis (10%, 3/33), persistent Mullerian duct syndrome (3%, 1/33), ovotesticular (3%, 1/33), Smith-Lemli-Opiz syndrome (3%, 1/33). Mutations in genes involved in gonadal development detected in 28% (17/60) patients, dominant mutations by frequency – AR (53%), SRY (17%), SF1 (12%), WT1 (6%), AMH (6%), DHCR7 (6%).**