DSD in Ukraine: our experience
Yuliya Shcherbak1, Nataliya Zelinska2, I. Schevchenko2, Evgenia Globa2
Anu Bashamboo3 and Ken McElreavey3

1 National Specialized Children Hospital “OKHMATDYT”, Center of Medical Genetics, Kyiv, Ukraine
2 Ukrainian Scientific Centre of Endocrine Surgery, Kyiv, Ukraine
3 Human Developmental Genetics, Institute Pasteur, Paris, France

The term “disorder of sex development” (DSD) includes congenital conditions in which development of chromosomal, gonadal or anatomic sex is atypical.

Methods
A retrospective analysis of the 75 medical cards of patients with DSD since 2000 up to 2017 years was done. Our database was created based on the self referring and does not cover all children with DSD in Ukraine. The criterion for including patients to the database was ambiguous genitalia and/or a discrepancy between the chromosomal and gonadal/genital sex.

At the time of examination the number of patients aged < 1 month was 17%, 1 month-up to 1 y.o. - 25%, 1-12 y.o. - 37%, >12 y.o. - 21%. The results of clinical data, laboratory tests and instrumental examination were analyzed. All patients (from birth to 18 y.o.) carried out a cytogenetic test, and, if necessary, fluorescence in situ hybridization (FISH). Molecular genetic testing was done in selected group of patients with 46,XY DSD in Ukraine (n=2) and in Institute Pasteur, France (n=19), using whole exome sequencing.

Results
In a group of patients with 46,XY DSD at first visit we suspected following clinical diagnoses:

| Androgen insufficiency syndrome | 3/18 |
| Perineal hypospadias | 7 |
| Complete gonadal dysgenesis | 6 |
| Bilateral anorhism | 5 |
| Androgen biosynthesis defect | 4 |
| Partial Gonadal dysgenesis | 3 |
| Ovotesticular DSD | 2 |

In a group of patients with 46,XX DSD we diagnosed: testicular 46,XX DSD (n=5), 21-hydroxylase deficiency with virilization IV-V degree by Prader (n=4), 46,XX gonadal dysgenesis (n=1) and DSD in VACTER-association (n=1).

Genetic testing in 46,XY DSD group was done in 21 (44%) cases. In 4 patients (19%) genes we found were not consistent with phenotype and their causality should be proven in further studies. In 4 (19%) patients no mutations were found.

In the entire cohort 6/75 (8%) cases the gender registration of the civil sex was changed during the first 2 years of life. A multi-disciplinary team has been created for gender assignment in DSD newborns and to improve the tactics of further management, including the time of gonadectomy.

Conclusions
Further studies to identify novel genes causing DSD are required.

References