**Difficulties in diagnosing variable disorders of sexual development**

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**Introduction:**
Disorders of sexual development (DSD) include etiologically heterogeneous group of patients that have disorders of genital development. Consensus guidelines that are currently used, divide all DSD in three main groups - sex chromosomal abnormalities, XX or XY DSD, all divided in subgroups in dependence of genetics and hormonal tests. The phenotypic spectrum of external genitalia, gonads and development of Wolfian and Mulerian duct derivatives varies in all patients. Many syndromic cases stayed unclassified and without easily reached etiology.

**Materials and methods**
14 patients with syndromic DSD were evaluated. All patients have ambiguous genitalia with different Prader staging. Phenotypic recognition, imaging, as well as karyotypic, hormonal and biochemical tests were evaluated in all. Excluded from the group: CAH, Turner sy, Klenefelter sy, isolated hypospadia, Swyer, AIS, Meyer-Rokitanski.

- 11 with XY karyotype, (3 SRY positive)
- 3 with XX karyotype, (2 SRY negative)

**Discussion and conclusion**
The diagnosis of DSD in the neonatal period represents one of the conditions that need urgent diagnosis and in some cases, early treatment. In some cases the condition stayed undetected till puberty. Clinicians often face many difficulties in performing and providing all necessary genetic and laboratory tests. Clinical workout and diagnostic evaluation paths were constructed in order to facilitate gender assignment in infants as soon as possible. Some of the investigations are not easily available, they are time-consuming, also some conditions still don’t have proven molecular defect. Advances in identification of the molecular and hormonal defect, as well as multidisciplinary approach improved the medical care, psycho social and ethical issues in patients with DSD.