Category:

P-18 **46,XX DSD with diphallus as a part of a multimalformative syndrome- a case raport**

E. Sukarova Angelovska E*, M. Kocova*, G Ilieva*, Anastasovska V*, Krstevska-Konstantinova M*, Filev G#

* University Children Hospital, Skopje, Macedonia

#University clinics of gynecology and obstetrics, Skopje, Macedonia

Disorders of sexual development (DSD) include etiologically heterogeneous group of patients that have disorders of genital development divided mainly in 3 groups- disturbances of sex chromosomes, gonadal dysgenesis or inappropriate hormonal action and synthesis that lead to disturbed phenotypic sex 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 hormonal tests . All conditions that are more complex, mainly stayed undiagnosed and part of a bigger picture are stated in the classification as other.

We present a case of a newborn where ambiguital genitalia is a part of the multimalformative syndrome. This was the first child in a family of unrelated parents. The pregnancy and delivery were uneventful. The baby had diphallus, and empty scrotum between. Examination revealed existance of Hirshprung anomaly, anal atresia and sacral agenesis. Analyses showed the female karyotype (46,XX), SRY negative, CAH screen negative. The baby had surgery, and died due to the complications, so more extensive tests were not performed.

There are more than 70 multimalformative syndromes associated with sexual ambiguity - either male or female phenotype and represent a separate group of DSD. A spectrum of other malformations includes skeletal, intestinal, renal, cardial system, accompanied with evident dysmorphism. Some of them have recognized chromosomal or molecular defect, but most of syndromic DSD's still don't have established mutation or pathway and are commonly recognized clinically by the continuum of associated anomalies. This DSD group should be included into the classification more precisely.

A systematic elucidation of
Differences of Sex Development

COST-Action BM1303 · www.dsdnet.eu

DSDnet

Certificate of Attendance

Elena Sukarova-Angelovska from **the former Yugoslav Republic of Macedonia**

attended the

International Workshop of Sex Development and Maturation

a collaboration of COST Actions
DSDnet, GNRH Network
and Endo-ERN

from

23.02.2018 to **24.02.2018**
in Lübeck (Germany)

Olaf Hiort
(Action Chair of DSDnet)

European Reference Network
for rare or low prevalence complex diseases