Introduction: Disorders of sex development (DSDs) constitute a group of congenital conditions that affect urogenital differentiation and are associated with chromosomal, gonadal and phenotypic sex abnormalities.

Objective: To evaluate the clinical and genetic features of childhood DSD cases.

Materials and Methods: DSD patients followed up between the years of 1981-2018 were included. The patients were evaluated in terms of their complaints, demographic, clinical features and genetic diagnoses.

RESULTS

46 XY DSD
- 47 (34.5%) of the patients had molecular diagnosis (Figure-3).
- 2 new suspected genes were detected by whole exome sequence analysis.
  - Homozygote c.332delC mut in CCDC60 gene
  - Homozygote c.36_41dupGGAGGC mut in ZNF653 gene
- Most common complaints of this group were hypospadias, undescended testis and ambiguty

46, XX DSD
- Pathogenic mutations was detected in 20 (33.8%) patients (Figure-3).
- Most common complaints were ambiguity, amenorrhea and cliteromegaly.

46 XX n:2
46 XY n:8
female
male

Figure 1: Distribution of DSD patients according to karyotype

Figure 2: Assigned gender of patients

Figure 3: Distribution of molecular diagnosis of 46 XY DSD and 46 XX DSD patients

Sex Chromosomal DSD
54 of the DSD patients were diagnosed as sex chromosomal disorder.
- 39 (72.2%) Turner Syndrome
- 3 (5.5%) Klinefelter Syndrome
- 10 (18.5%) Mixed gonadal dysgenesis
  - 1 (1.8%) 47 XXX
  - 1 (1.8%) 47 XYY

Figure 4: Gender change n:10

*Genetic diagnosis is still not available in many of the DSDs that have emerged with complex genetic mechanisms

*Many patients undergo erroneous gender determination prior to admission to endocrine centers

*A multidisciplinary approach is needed in the process from diagnosis to gender selection and follow-up