Clinical and Molecular Spectrum of a of Patients with Disorders of Sex Development: A Single Center Experience

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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

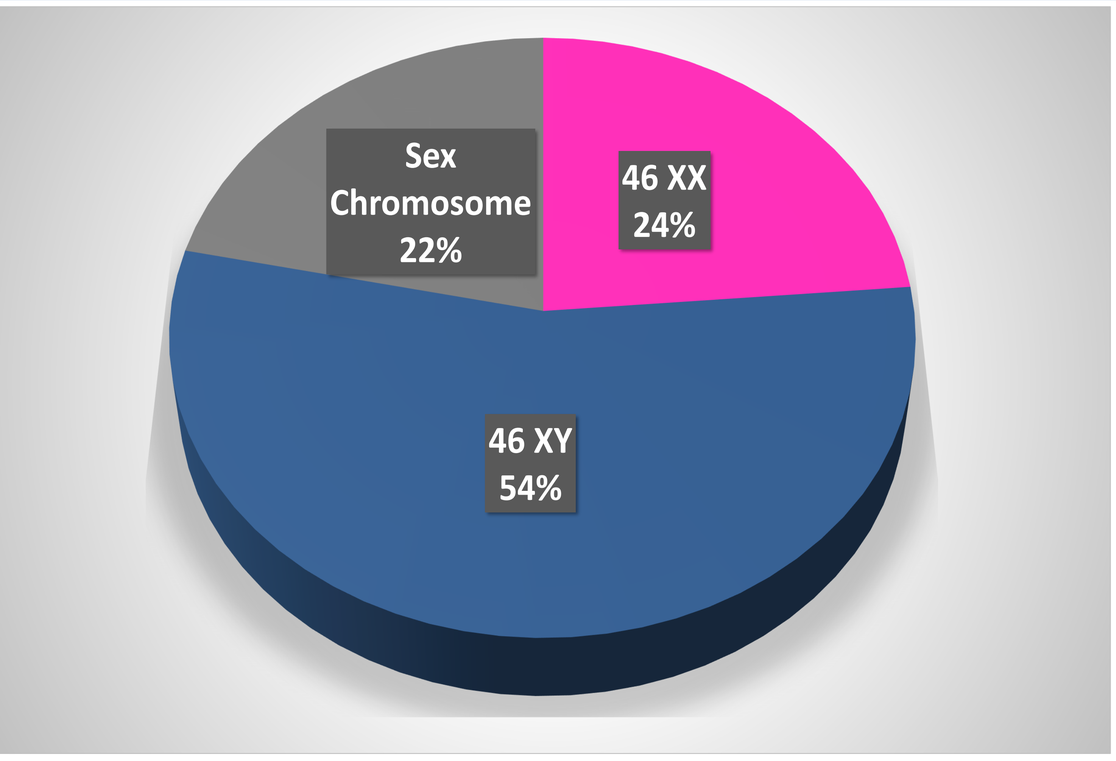


Figure 1: Distrubution of DSD patients according to karyotype

250 patients;

- Median age at admission was 5,2 (0-19) years.
- Molecular diagnoses was made in 121 of the patients (48%).
- Overall 39,9 % of the patients were born from a consanguineous marriage.

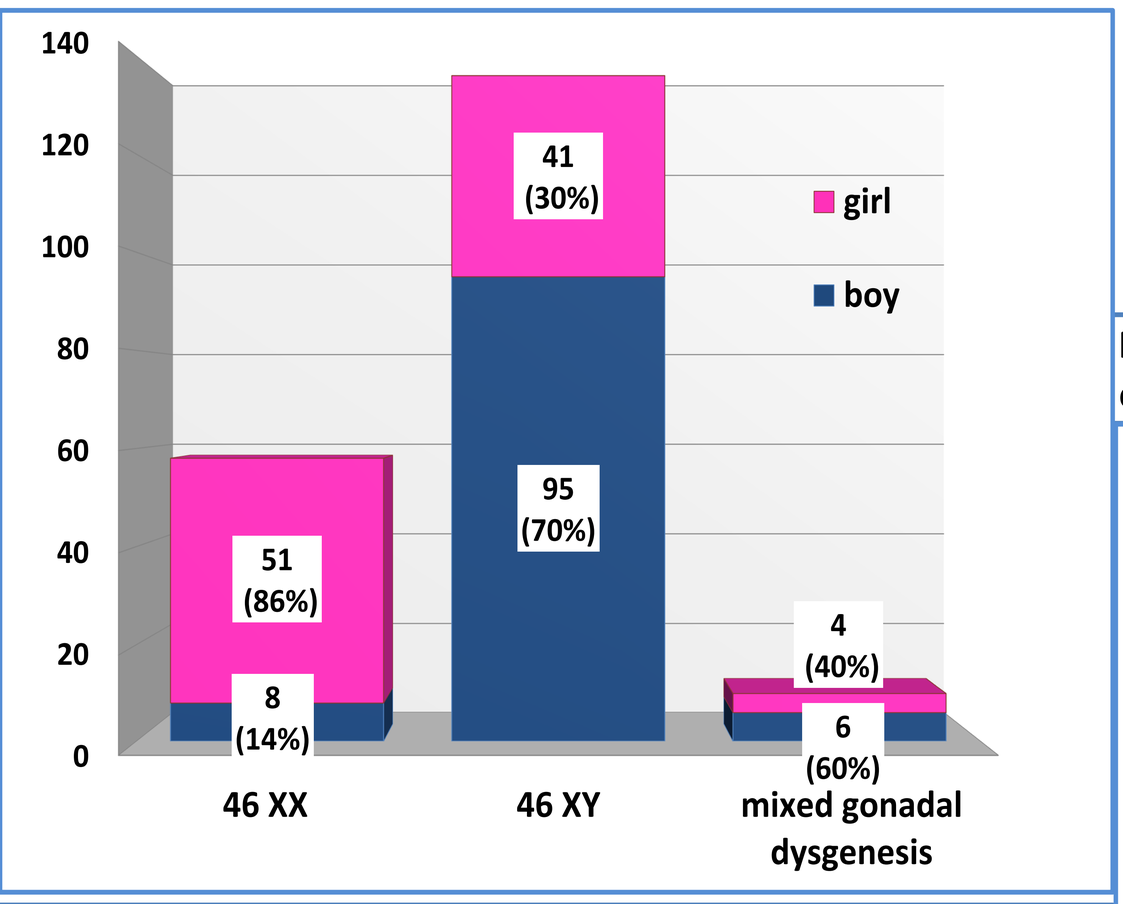
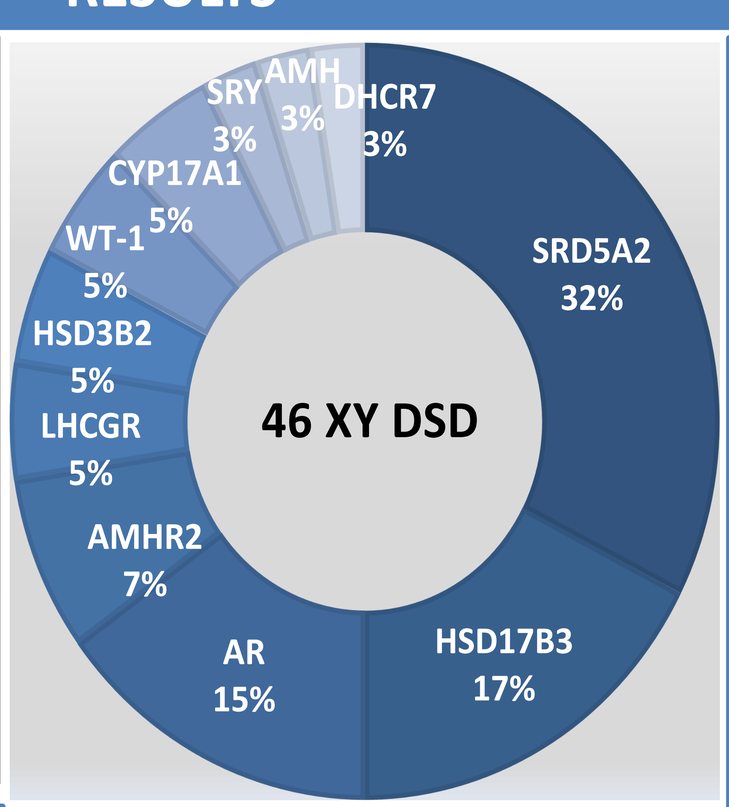


Figure 2: Assigned gender of patients



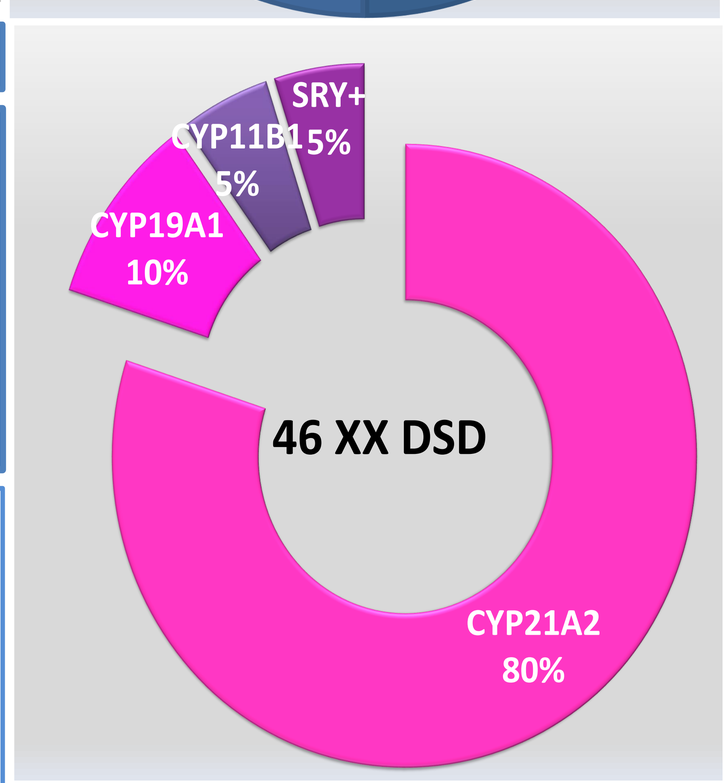


Figure 3: Distribution of molecular diagnosis of 46 XY DSD and 46 XX DSD patients

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

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 ambougity

46, XX DSD

- Pathogenic mutations was detected in 20 (33,8%) patients (Figure-3).
- Most common complaints were ambougity, amenorrhea and cliteromegaly.

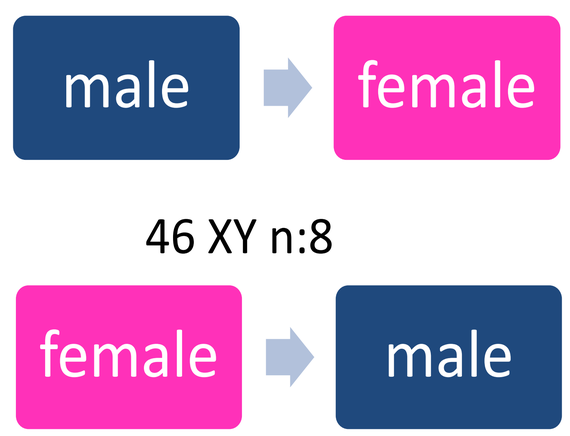


Figure 4: Gender change n:10

*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