

SEX ASSIGNMENT AT BIRTH : IS IT ALWAYS EASY ?

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INTRODUCTION

Disorders of sex development (DSD) are a group of congenital disorders in which the development of chromosomal, gonadal and anatomical sex is atypical and disharmonious. These DSD may constitute a medical emergency (salt wasting syndrome in congenital adrenal hyperplasia) or a social emergency due to the parents' anxiety generated by the difficulty of assigning the child's sex at birth. Providing an urgent and adapted care defining the sexual orientation of the child remains challenging.

AIM

The objective of this work was to describe the clinical, diagnostic and therapeutic aspects of DSD at the department of Endocrinology-Diabetology and Nutrition of Mohammed-VI University Hospital, Oujda, in the eastern of Morocco.

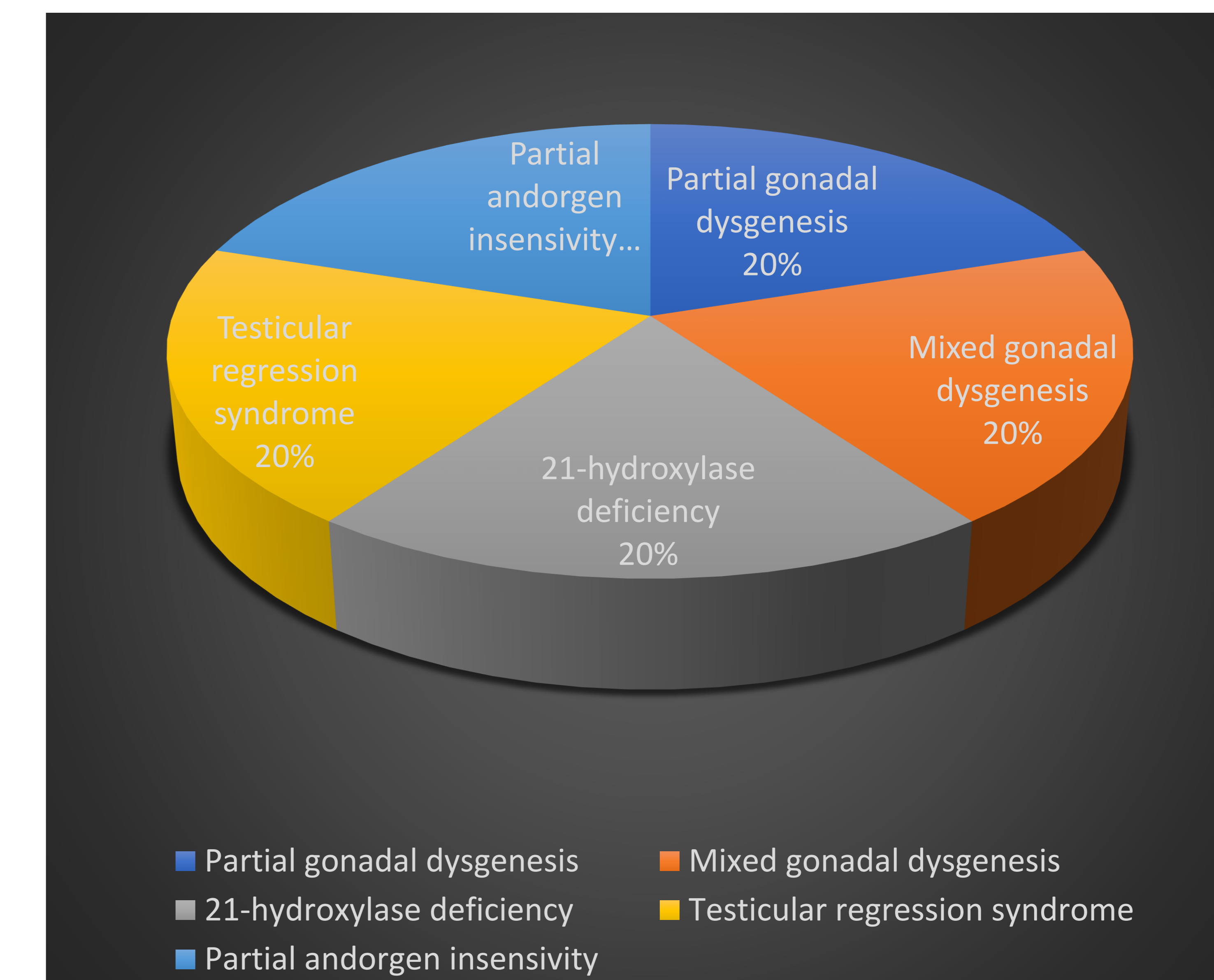
METHOD

Descriptive, cross-sectional study of 24 patients followed at the Mohammed-VI University Hospital Center of Oujda for defects in sexual development.

RESULTS

The average age was 6.2±4.5 years. These patients were from a consanguineous marriage in 22% of cases. A delay of sex assignment at birth was found in 55.6%. The diagnosis of a defect in sexual development was made in the neonatal period in 55.6% of cases and in the pubertal period in 11.1% of cases. Sixty-six percent of the patients had external genitalia abnormalities and 22.2% had external genitalia defects associated with a growth delay. Genetic evaluation revealed a male chromosome formula (46, XY) in 65.5% and a female chromosome formula (46, XX) in 22.2%. A mosaic chromosome formula with a double cell population was found in 12.3%. The final diagnosis was partial gonadal dysgenesis in 22.2% of cases, mixed gonadal dysgenesis in 22.2%, 21-hydroxylase deficiency in 22.2%, testicular regression syndrome in 22.2% and partial androgen insensitivity in 11.1%.

The therapeutic management was multidisciplinary. Feminizing genitoplasty was performed in 22.2% of cases, hypospadias was cured in 33.3%, micropenis was corrected by dihydrotestosterone in 66.7% and gonadectomy was performed in 22.2% of patients. Patients with classic form of 21-hydroxylase deficiency were treated with hydrocortisone and fudrocortisone. GH substitution was used in 22.2% of cases. Psychological support was offered to all patients according to their age and to all their relatives.



CONCLUSIONS

DSD is a rather rare entity. The diagnosis is generally made during the neonatal period when abnormalities of external genitalia are observed. These pathologies require a rigorous diagnostic approach in which the blood karyotype plays a key role in order to distinguish between 46, XX DSD; 46, XY DSD patients and those with gonosomal abnormalities. Their management should be multidisciplinary.

