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(CASE REPORT)

46 XY disorder of sex development: Case report

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Abstract

Disorders of sexual differentiation in 46, XY patients cover a wide spectrum of clinical pictures (female phenotype, posterior hypospadias, cryptorchidism). Their causes are varied and affect the stages of testicular determination (gonadal dysgenesis) or defects in testosterone production or action (testosterone secretion abnormalities, androgen insensitivity, 5α-reductase deficiency). They require rational and rapid management, within a multidisciplinary team. We report a rare case of this disorder in a patient of 18 years with the therapeutic obstacles that we faced in its management.

Keywords: 46 XY DSD; Chromosomal sex; Phenotypic sex; Multidisciplinary approach

1. Introduction

Disorder of sexual development (DSD) is referring to congenital conditions in which development of chromosomal, gonadal or anatomical sex is atypical. A new classification of DSD includes three major groups: sex chromosome DSD, 46, XX DSD (disorders of ovarian development, androgen excess and others) 46, XY DSD (disorders of testicular development, disorders in androgen synthesis or action and others). the term "46, XY DSD" is characterized by the presence of 46, XY karyotype, exclusively male gonads and ambiguous or female external and/or internal genitalia, caused by incomplete virilization during prenatal life. These disorders can arise from a variety of conditions, including dysgenesis of the gonads during embryological development, abnormalities of gonadotropins, inborn errors of testosterone biosynthesis and many abnormalities of androgen target cells. The clinical management is complex. A multidisciplinary approach to this problem involving endocrinologists, specialists in the field of genetics, surgery and psychiatry is necessary in order to reach a prompt and correct diagnosis and treatment.

2. Case report

A 17-year-old female patient, without particular pathological history, consulted for a primary amenorrhea. The interrogation was without particularities. the clinical examination objectified signs of hyper androgenism, in particular a light Hirsutism rated at 15 according to the score of Ferriman and Gallwey, frontal gulfs, masculine musculature and acne of the upper face of the thorax.

The genital examination showed male external genital organs with a 4.5 cm penis size and 4th stage of PRADER. (figure 1 ,2).

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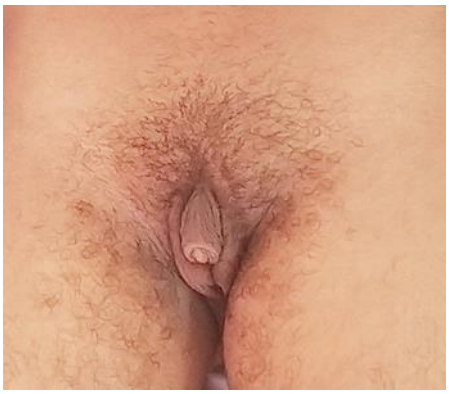

Figure 1 Genital anatomy

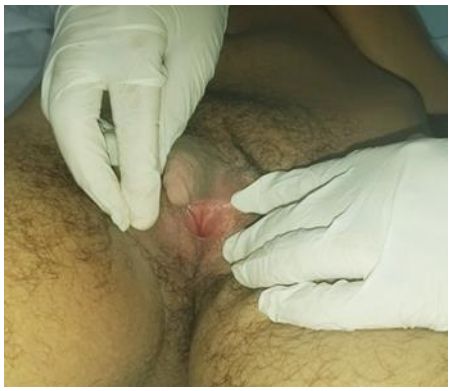

Figure 2 Genital anatomy

After an accurate physical examination, we decided to perform several investigations:

- Hormonal dosage revealed a low level of estradiol 18,8 pg/ml , a Hight level of testosterone 4,9 ug/l.
- Pelvic ultrasound (US) showed the absence of uterus and ovaries; Presence of small bilateral oval images under the bladder with small cystic areas that may be related to seminal vesicles.
- Pelvic MRI: Individualization of two tubular structures simulating seminal vesicles on either side of a small median nodular structure simulating a prostate; Evidence of two testicles at bilateral inguinal level measuring 26*13mm on the left and 31*12mm on the right; Hypoplastic penis
- According to the genetic evaluation, we performed the karyotype analysis that showed normal male karyotype (46,XY).
- A psychiatric expert recommended that the patient's female gender should be respected.

Our decision was to respect the female sex of our patient and to make a bilateral gonadectomy with a genitoplasty before starting a hormone replacement therapy, except that the legislation of our country prohibits any procedure aiming to change the gender.

3. Discussion

The term DSD refers to disorders that affect the normal process of sexual development causing disagreement between chromosomal, gonadal and phenotypic sex. The incidence of DSDs in live births is estimated to be in the range of 1/4,500 to 1/5,000 (1). Patients with DSDs present with diverse clinical manifestations. In some patients, genital ambiguity is apparent at birth, whereas others have typical male or female genitalia at birth but a different internal gonad structure, which can delay clinical diagnosis until adolescence or adulthood (2).

Individuals with 46, XY karyotype and disagreement between external genitalia and gonadal sex are classified as individuals with 46, XY disorders of sexual development (DSD). Most of these patients have an autosomal recessive pattern of inheritance linked to X chromosome (3).

Patients with 46, XY DSD have lower virilization of genitals compared to normal 46, XY individuals. Etiology may be associated to hypoplasia of Leydig cells, enzyme disturbances of testosterone synthesis, deficit of 5-alfa-reductase enzyme (DEF5α), testicular regression syndrome, gonadal dysgenesis (GD), androgen insensitivity syndrome (AIS) or ovotesticular DSD (4)

Once the diagnosis is established, other pivotal step in 46XY DSD management is the assignment of sex. This decision should consider that gender identity often disagrees with genetic, gonadal, or anatomic sex and that sexual attraction is challenging to predict according to sex phenotype. Firstly, it is crucial to gather a multidisciplinary team, including endocrinologist, urologist, geneticist, radiologist, and mental- health specialist, to avoid distress Thus, a straightforward and clear explanation about development, fertility, sexual function, and need for medical/surgical intervention must be given to the patient before deciding the sex (5)

Before deciding on the sex of rearing, it is also fundamental to consider condition specific outcomes related to psychosexual development, anatomy, fertility potential, and the need for medical and/or surgical treatment. Finally, patient cultural and religious beliefs must be respected when deciding gender assignment. At this point, another critical component for 46 XY DSD management is hormone replacement. The goals for this treatment are the induction and maintenance of secondary sex characteristics, growth, bone mineralization, and ameliorating psychosocial and psychosexual development and general well-being. Induction and maintenance of puberty are essential despite the gender identity.

4. Conclusion

Disorders of sexual development are a rare entity. The complexity of the problem requires a multidisciplinary team working together. Affected patients and their parents should be provided with full information to make an appropriate choice for gender assignment.

Compliance with ethical standards

Acknowledgments

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Disclosure of conflict of interest

No conflict of interest.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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