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CASE REPORT

Post-pubertal presentation of 46XY disorders of sex development: A case report

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INTRODUCTION

Disorders of sex development or differentiation (DSD) are congenital conditions that arise from chromosomal, gonadal and phenotypic sex abnormalities It is classified into three types according to chromosomal abnormality, such as 46XX DSD, 46XY DSD and sex chromosomal DSD.[1] These abnormalities occur due to a complex process at any point in genetic interaction, androgen synthesis and interaction of ligands with corresponding receptors. The incidence of 46XY DSD is 1 in 20,000 live births. Its diagnosis may occur at various ages and the etiolopathogenesis may not be found in many cases.[2] In patients with this abnormality, the sex of rearing poses great importance for proper gender identification. An individual's psychological development depends on the interaction of genetic makeup, hormonal aspects, and sex of rearing. When this harmony breaks, the individual becomes socially unacceptable. In these cases, the decision of gender assignment is based on the patient's potential to live in the desired gender, psychological adjustment after surgery, quality of life, and social and cultural acceptance.[3] Proper counseling of the patient and family regarding the timing and type of surgery and their pros and cons are needed.[4] There are very few case reports worldwide and no such case report like this in Bangladesh. Here we present a case of post-pubertal presentation of a 46XY DSD patient who was reared up as a girl by her parents and well accepted in society.

LEARNING POINTS

1. Social awareness, of all concerned are needed to break the taboo about individuals with ambiguous genitalia.
2. Early diagnosis of 46XY, disorders of sex development or differentiation and management should be done involving urologist, pediatrician, endocrinologist, gynecologist and psychiatrist that will ensure appropriate sex of rearing according to genetic sex and to promote a socially compatible good quality of life.

CASE DESCRIPTION AND MANAGEMENT

A 26-year-old female graduate service holder sought medical attention at a local hospital with primary amenorrhea, lack of breast development and a phallus-like growth since birth. Her parents raised her as a female. She visited the Endocrinology Department of Bangabandhu Sheikh Mujib Medical University (BSMMU). Upon physical examination, she had severe penoscrotal hypospadias with a bifid scrotum looking like labia containing testis, stretched penile length of six cm, and a blind vaginal pouch. Her tanner score for breast was one, pubic hair was four, and she had facial hair. Her karyotype showed 46XY, indicating male. Hormonal evaluation revealed that her serum testosterone, luteinizing hormone, prolactin, adrenocorticotropic hormone, and basal cortisol were all within the normal limit. However, due to resource limitations serum dihydrotestosterone, T:dihydro-testosterone, β-hcg stimulation test and genetic testing couldn't be done. Ultrasonogram of whole abdomen revealed no prostate or uterus. But contrast magnetic resonance imaging of the whole abdomen revealed a

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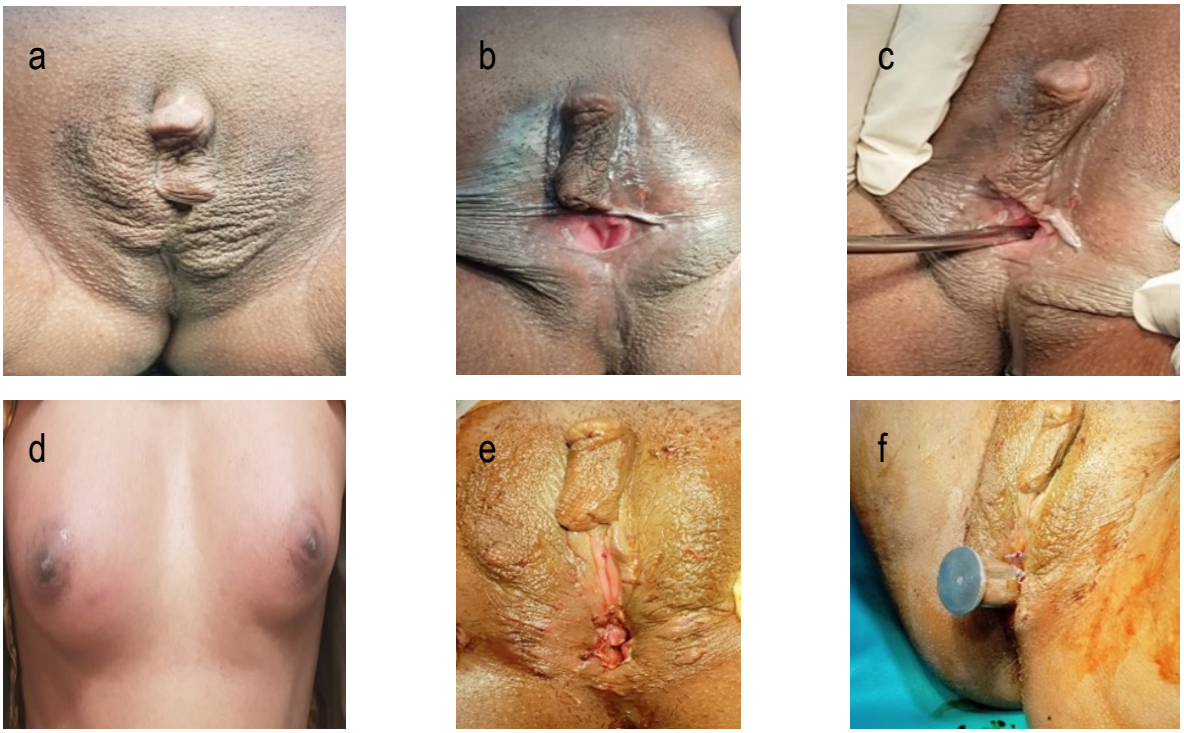

FIGURE 1 **All are preoperative pictures before vaginoplasty. a) showing external genitalia of the patient having small phallus, bifid scrotum. b) showing small vaginal pit. c) showing external urethral meatus allowing passage of Hegar's dilator suggesting penoscrotal hypospadias. After about two years of estrogen therapy, the patient developed d) well-developed both-sided female breast and e) showing external genitalia after vaginoplasty and f) showing neovagina permitting 9Fr anal dilator almost full length suggesting adequate caliber and length of the neovagina.**

small prostate but no uterus or ovary. She was stimulated to ejaculate, and the semen was found to be normal according to WHO criteria. Subsequently, she was referred to the Female Urology and Andrology division of the Department of Urology, BSMMU with a diagnosis of 46, XY DSD for possible surgical reconstruction.

Proper counseling and shared decisions were made with the patient and her family regarding genital reconstruction. Since she was already socially accepted as a female and had sexual attraction towards males, she decided to remain female. A multidisciplinary approach involving an endocrinologist, psychiatrist, and gynecologist was taken to make the decision.

As per the decisions made, she underwent bilateral orchidectomy with penectomy and clitoral reconstruction in the first setting in March 2022. She started receiving estrogen as hormone replacement therapy for her breast development and laser treatment for facial hair.

After about two years, she presented for her second-stage surgery. Upon examination, her external genitalia looked like a female with near normal clitoris and labia majora with blind ending vaginal pit and reduced facial hair, tanner score for breasts two and pubic hair of five. Proper counseling and consent were taken from her and her future partner (as she was engaged) and family about future conjugal life and fertility issues after surgery. Then she underwent Vaginoplasty by abdominoperineal route using a peritoneal flap. The vaginal mold was removed and the neovaginal canal permitted 9Fr anal dilator completely. She was advised to continue regular vaginal dilation with a wider dilator to attain girth of a normal erected penis.

DISCUSSION

To diagnose a DSD, a detailed history and physical examination including genital and inguinoscrotal region is needed. Physical examination reveals ambiguous genitalia. Karyotyping is needed to confirm the sex chromosome. Additionally, hormonal assessment,

imaging and genetic testing is necessary. The treatment options include surgery, hormone therapy and psychosocial therapy. The surgical options include gonadectomy followed by clitoroplasty, labioplasty and vaginoplasty.[5]

A previous case study from India showed, a 13-year-old girl presented with primary amenorrhea, ambiguous genitalia and lack of breast development. After a thorough evaluation 46XY DSD was diagnosed. She underwent clitoroplasty, bilateral gonadectomy and vaginoplasty followed by estrogen therapy.[6]

Another case of a 44-year-old 46XY DSD patient from Vietnam presented with abnormal genitalia and primary amenorrhea. She underwent breast augmentation, penectomy, orchiectomy, the reconstruction of clitoris, labia and vagina.[7]

In Indonesia, three siblings at post-pubertal age, were diagnosed as 46 XY DSD in 2022, reared up as female. One of them chose to remain female and underwent clitoroplasty, orchidectomy, estrogen supplementation and vaginoplasty.[2, 8] In such cases, the surgery should offer the patient the best prospect for healthy puberty and sex life.[9] Moral support from family members and specialists is needed to alleviate the fear and boost psychologically.[10]

As 46 XY DSD is a rare disease, a high degree of suspicion is needed to diagnose the condition early for proper management which will ensure proper sex of rearing and better quality of life to the patient. Extensive efforts should be made to break the taboo. Extensive research and social awareness are needed to break the taboo regarding DSD.

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Author contributions

Manuscript drafting and revising it critically: MS, MSI, MNI. *Approval of the final version of the manuscript:* MS, MSI, MNI. *Guarantor of accuracy and integrity of the work:* MS, MSI, MNI.

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Conflict of interest

We do not have any conflict of interest.

Ethical approval

Ethical approval was not sought because this is a case report. However, informed written consent was obtained from the patient for preparation of this manuscript.

Data availability statement

The data that support the findings of this study are available on request from the corresponding author.

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