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Case report

Vulvar-vaginal reconstruction surgery for sexual function in patient with disorders of sex development

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ABSTRACT

Introduction and importance: Amenorrhea can be a transient, intermittent, or permanent condition reflecting the overall health condition of a woman. Primary amenorrhea and ambiguous genitalia appearance warrant more comprehensive diagnosis and treatment, especially in those planning to have a married life.
Case presentation: A case a 24 years old woman was referred to our hospital with a chief complaint of primary amenorrhea. Previously, she was diagnosed with vaginal agenesis. However, a thorough examination revealed ambiguous genitalia and karyotype result of 46, XY. Following several rounds of discussion with the patient, her family, and her partner, she chose to remain a female and even planned to be married following the treatments. A vaginoplasty followed by clitoral reduction was done with a satisfactory result.
Clinical discussion: Disorders of sex development and differentiation is a rare but devastating condition. Various biological, psychological, and social issues are surrounding those affected by it. The burden was further aggravated for those who are late to discover their condition, especially those planning to be married.
Conclusion: Comprehensive and multidisciplinary approaches are needed to ensure the best outcome for DSD patients.

1. Introduction

The menstrual pattern can be a reliable indicator of the overall health status and self-perception of the well-being of a woman. [1] Amenorrhea may be a transient, intermittent, or even permanent condition with various etiologies, ranging from pregnancy to congenital problems. Primary amenorrhea, absence of menarche with no previous history of menses, should prompt a thorough evaluation to determine the cause. [2] One of these causes may be disorders of sex development.

Disorders of sexual development (DSD) requires a precise, timely, and complicated organization of various interdependent factors. Should any of the cooperating factors fail, a person would be afflicted with disorders of sex development. This is a devastating condition with various complications. [3,4]

In this study we would like to present an atypical case of a woman with primary amenorrhea, who was after a thorough examination, had 46, XY karyotype and disorders of sex development. The case report was made in accordance with the SCARE 2020 guideline. [5]

2. Case presentation

A 24 years old woman was referred to our clinic with a chief complaint of primary amenorrhea. She realized that she was not menstruating when she was seventeen. She paid no attention as there was no additional complaint. She had only realized the issue after she prepared to marry her current partner. Previously, she was evaluated by an obstetrician with the result of a small uterus and the absence of a uterus. However, her external genitalia was deemed ambiguous, thus she was referred to our center.

Other than the primary amenorrhea, she had other complaints such as urinating or defecating problems. There was no history of previous illness before the visit. Moreover, there was no similar problem in her family. She was the fifth child in her family, with all of her brothers and sisters already married and having children. During the initial

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examination, her height was 170 cm with a body weight of 63 kg. Her vital signs were also within normal limits. Her tanner stage was M1P3.

During external genitalia examination, it was revealed that she had ambiguous genitalia, with clitoromegaly resembling a penis (size 1 cm × 1 cm × 1.5 cm) and no visible vaginal introitus. Her pubic hair growth and spread were within normal limits. There was no palpable vaginal structure recognized during the rectal toucher examination. Her anal sphincter tone was fine with smooth rectal mucosa. (Fig. 1).

During a genital ultrasound examination, it was revealed that there was no scrotal structure in the vulvar region, and there was no testicular structure in the labial, inguinal, or pelvic area. There was also no uterus visualized during the examination. Furthermore, a whole abdomen magnetic resonance imaging (MRI) examination revealed no structure resembling the uterus and adnexa. There was a structure resembling testes bilaterally at the inguinal region. (Fig. 2). Following the examination, a prompt laboratory and genetic examination were performed.

Her laboratory result was within normal limits. However, her karyotype result showed that she had 46, XY karyotype. A multidisciplinary DSD team of urogynecologists, urologists, forensic psychiatrists, endocrinologists, and genetic experts was assembled to discuss the condition with the patient, her partner, and her family. She was diagnosed as having complete androgen insensitivity syndrome. After several rounds of counseling, the patient chose to become a woman. Her partner and she chose to accept her condition and also planned to marry following the treatments. During the treatment, hormonal therapy using progesterone, bilateral orchidectomy, vaginal introitus widening surgery with Fenton's procedure to create a functional vagina (Fig. 3), and a clitoral reduction surgery using a nerve-sparing technique (Fig. 4) were done for her by first author and team. The bilateral orchidectomy was done by our urologists.. The patient was discharged two days following the surgery.

Her bilateral gonads histopathology results showed dysgenetic testis appearance, with both the left and right testicle appearing to have normal testicular structures (epididymal ducts, Sertoli cells, vas deferens) but atrophic seminiferous tubules with interstitial fibrosis and no spermatogenesis observed.

A month following the surgery, the patient and her partner came for a follow-up examination. They told the clinicians that their sexual intercourses were rather satisfactory, albeit having to use artificial lubricants for their intercourse. She also did not have any complaints regarding her surgery result.

3. Discussions

Primary amenorrhea is an alarming symptom prompting more thorough evaluation to identify the cause. Usually, primary amenorrhea is diagnosed when menses were absent at the age of 15 years old in the presence of normal growth and secondary sexual characteristics. [2,6] Thus, patients complaining of primary amenorrhea are usually present at pubertal and adolescent ages. Usually being a result of genetic or anatomical abnormality, primary amenorrhea should be thoroughly evaluated no matter the age of the patient.

Detailed history regarding menstrual patterns, eating and exercise habits, psychosocial stressors, medication, chronic illness, breast, and pubic hair development, or other complaints should be recorded. Physical examination of height, weight, body mass index, breast development, and genital appearance should also be observed. Moreover, additional examinations such as pregnancy tests, serum hormones, pelvic ultrasonography, and karyotyping may be necessary. [1,2]

A study by Klein et al. (2019) categorizes primary amenorrhea causes as outflow tract abnormalities, primary ovarian insufficiency, hypothalamic or pituitary disorders, endocrine gland disorders, sequelae of chronic disease, physiologic, or induced amenorrhea. [1] The most common etiologies of primary amenorrhea include gonadal dysgenesis, Mullerian agenesis, physiologic delay of puberty, polycystic ovary syndrome, hypopituitarism, and anorexia nervosa. [7] However, rare cases of primary amenorrhea include complete androgen insensitivity syndrome, a form of disorders of sex development (DSD). The patient in our case was also diagnosed as having complete androgen insensitivity syndrome, a rare and often overlooked syndrome.

Disorders of sex development (DSD) are a myriad of congenital conditions causing atypical development of chromosomal, gonadal, and anatomic sex. It is usually grouped in accordance with the result of karyotype and additional examinations into 46XX, 46XY, sex chromosome, ovotesticular, and non-hormonal DSD groups. [8] Due to the limited evidence regarding DSD, diagnosis, and management of DSD remains a challenge to both clinicians and researchers worldwide. [8,9]

The problems affecting DSD patients are not only limited to biological problems, but also psychological, social, and even ethical problems. The feeling of losing identity as either male or female, alienation from society because of being seen as different, and choosing a gender would be overwhelming for most patients with DSD. [9–11] Therefore, it is also thought of as both a medical and social emergency. In our case, the

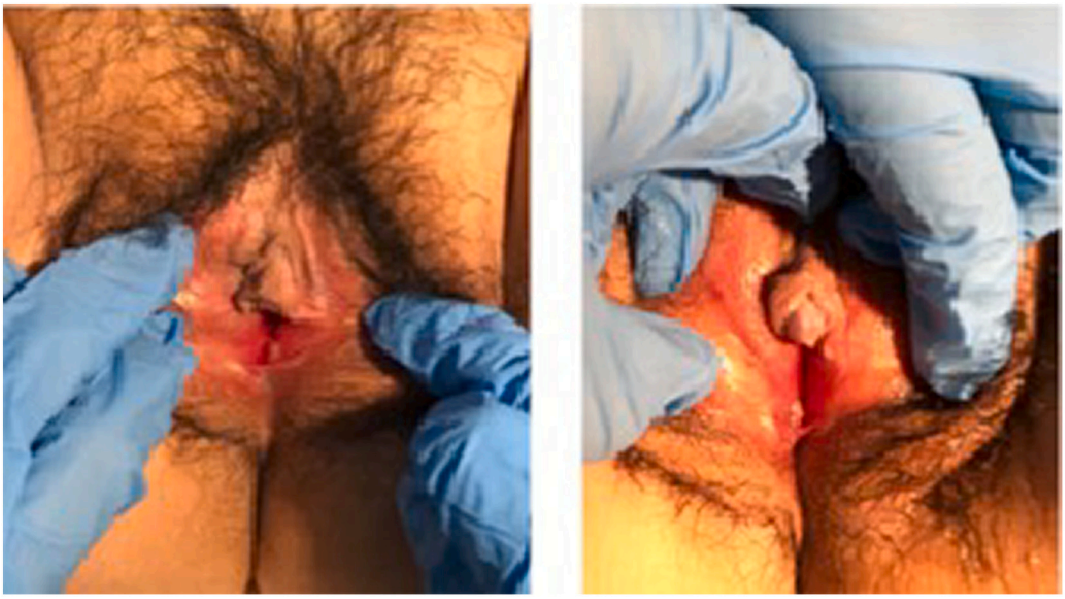

Fig. 1. External genitalia appearance of the patient.

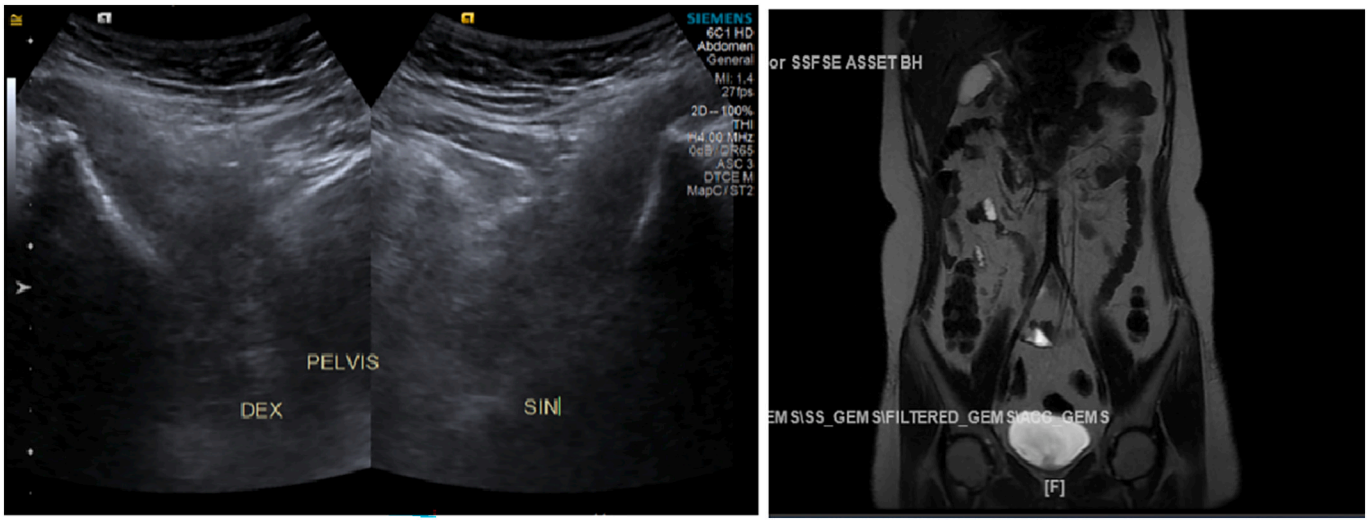

Fig. 2. Ultrasound and Magnetic Resonance Imaging (MRI) examination results showed no signs of uterus or vagina.

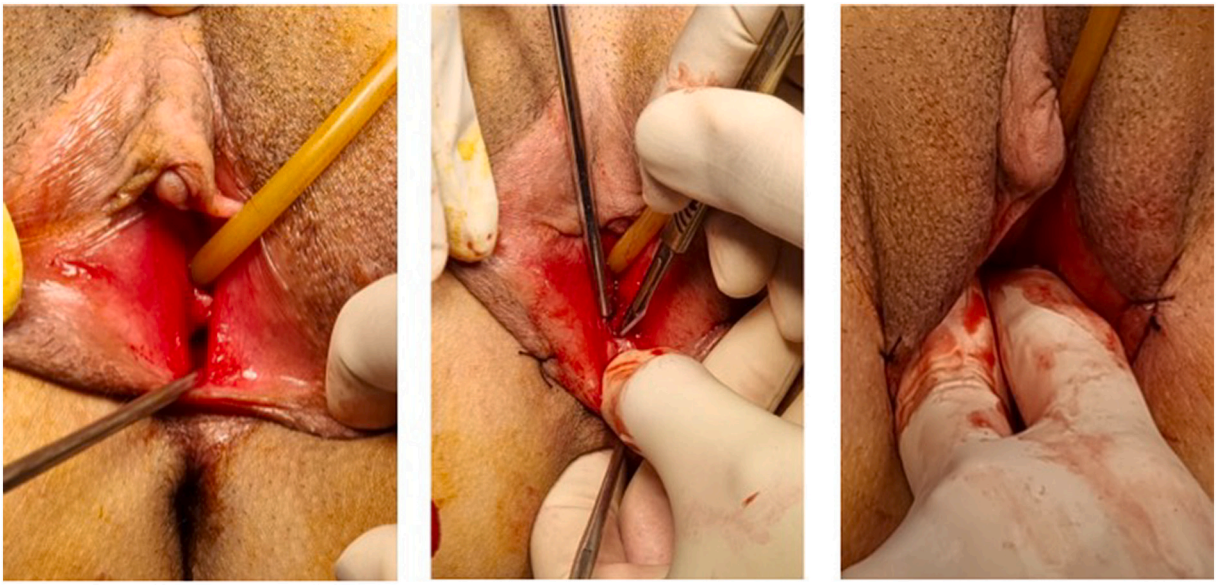

Fig. 3. Vaginal introitus widening with finger and speculum and Fenton's procedure.

problem was even more severe as the patient had already established her gender identity and even planned to be married. Fortunately, after several rounds of consultations and discussions, she accepted her condition and had also chosen to remain a female.

Vulvar-Vaginal reconstruction and clitoral reduction in our case were done following the gender assignment of the patient. After planning to have a marriage as a woman, hormonal therapy using progesterone, bilateral orchidectomy, and vaginal introitus widening surgery were performed to create a functional vagina for the patient. [4] In order to avoid using busination for creating neovagina for the patient, the vaginal introitus widening was done with the help of a finger and speculum. Meanwhile, clitoral reduction in our case was done in a nerve-sparing manner to conserve sexual satisfaction. The overall result and sexual satisfaction following the surgery were satisfactory. [12]

After the gender assignment and reconstructive surgery of patients' genitalia, there are other follow-up actions that should be done. Assimilation to the families and previous social circles, reintroduction as a new gender, and psychiatric observations should be performed to ensure the best outcome following the treatments for DSD patients.

Currently, studies regarding DSD patients are limited to case reports and small studies. We have known that DSD patients are faced with various biological, psychological, and social problems. However, there are special cases needing more attention, such as those discovering their condition late, even only after having a plan to marry. This study was made to elucidate the scope of the problem with lately discovered DSD and the management needed to ensure the best outcome for the patient.

4. Conclusion

Primary amenorrhea is an alarming symptom prompting more thorough examinations as it may become a chief complaint of the more severe problems surrounding the condition, such as in DSD patients. A comprehensive and multidisciplinary approach is needed to ensure the best outcome for the patient.

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The study was fully funded by the authors.

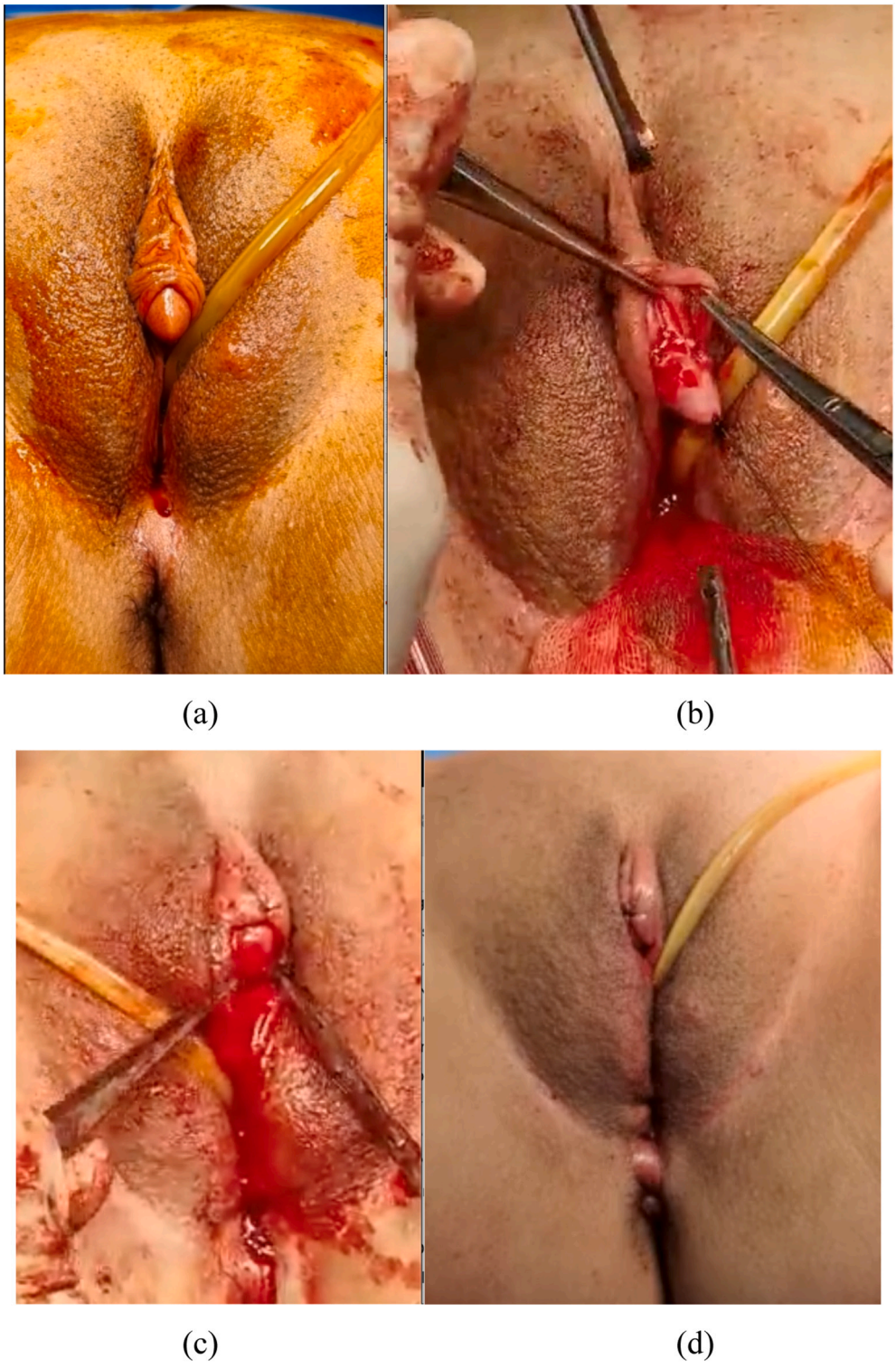

Fig. 4. Nerve-sparing clitoral reduction (a) initial appearance (b) excision of excess skin (c) reduction and relocation of clitoris (d) final appearance.

Ethical approval

The ethical approval for this study was published by the Medical Research Ethics Committee, Faculty of Medicine, Universitas Indonesia, Indonesia.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Research registration

Not applicable.

Guarantor

The corresponding author would like to be the guarantor for this study.

Provenance and peer review

Not commissioned, externally peer-reviewed.

CRediT authorship contribution statement

FM was involved in conceptualization, protocol development, administration of the study, data collection, funding, manuscript writing, and manuscript finalization.

DN involved in conceptualization, protocol development, administration of the study, data collection, and manuscript finalization.

SH involved in conceptualization, protocol development, administration of the study, data collection, and manuscript finalization.

TP involved in protocol development, study conceptualization, patient recruitment, and AFP editing and finalization.

APM contributed in conceptualization, administration of the study, funding, data collection, and manuscript editing and finalization.

GNH contributed in conceptualization, protocol development, data collection, and manuscript finalization.

Declaration of competing interest

There was no conflict of interest to be disclosed.

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