Dr. Sulaiman Al Habib Medical Journal
Vol. **1**(**3-4**); December (2019), *pp.* 70–72
DOI: https://doi.org/10.2991/dsahmj.k.191118.001; eISSN 2590-3349
https://www.atlantis-press.com/journals/dsahmj

Case Report

Sexual Ambiguity—A Social Predicament

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ARTICLE INFO

Article History

Received 22 September 2019
Accepted 30 October 2019

Keywords

Ambiguous genitalia
disorders/differences of sex development
ethics
decision-making
social dilemma

ABSTRACT

Among the diverse clinical manifestations of Disorders of Sex Development (DSD), the ambiguous look of genitalia is the most typical disorder. In this condition, the sex of the newborn cannot be readily differentiated because of atypical appearance of the external genitalia. Thus, the treatment of such a child is composite from both sociocultural and medical viewpoints. Decisions that affect the future sexual function and sex assignment of the child are made on behalf of the child by their parents and healthcare professionals. Such decisions are rarely simple and straightforward. The outcome data resulting from such decisions is limited and conflicting, and cannot offer clear guidelines to the community. This case presents a 20-year-old patient who presented with primary amenorrhea and delayed pubertal features, and was diagnosed as 46, XY typical male genotype. As she had presented with a female phenotype at birth, she had been raised as a female and developed a consistently female sex identity. She underwent surgery, during which remnant testes were removed, and bilateral breast augmentation and clitoroplasty were performed. This decision to be rehabilitated as a girl was made by the patient herself. In this part of the world, sexual variation that blurs the line between male and female is stigmatized. The birth of a child with DSD is emotionally traumatizing and considered a social stigma and taboo. Therefore, social familiarity regarding this treatable disorder is currently needed in Pakistan irrespective of the cultural, social, and religious beliefs, and to protect the society from the ambiguous obscurity of the unthinkable of what was earlier called "intersex disorders."

1. INTRODUCTION

Disorders of Sex Development (DSD) is an umbrella term for a large number of rare conditions on atypical sex development, leading to sexual ambiguity and infertility. DSD is defined by congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical [1]. In this condition, the neonate external genitalia do not emerge to be illustrative (whether male or female) [2].

It is estimated that the incidence of all types of DSD is between one in 2500 and one in 4500. The incidence of Congenital Adrenal Hyperplasia (CAH; a specific type of DSD) is about one in 15,000 in most countries. Many types of DSD have autosomal inheritance; hence, in communities in which consanguine marriages are common, DSD is more frequently seen [1]. The incidence of ambiguous genitalia in Saudi Arabia is shown as one in 2500 newly born and in Egypt one in 3000 live births [1]. However, the incidence of ambiguous genitalia in Germany is two per 10,000 live births [3], whereas in the UK it is about one in 18,000 live births. There is shortage of data regarding incidences of ambiguous genitalia in developing countries. Most of these cases have been reported from Western societies [4].

There are different causes of ambiguous genitalia in newborn, but the most common causes in inherent females are CAH, maternal ingestion of substances with male hormone activity during gestation, and male hormones generating tumor in female fetus, whereas possible causes in males genome are Leydig cell aplasia, 5-alpha-reductase deficiency, androgen insensitivity syndrome, and maternal ingestion of substances with female hormone activity during pregnancy [4–6].

Worldwide, DSD is commonly treated as a taboo, in which newborns with this disorder are hidden from the society because of shyness and coyness. In our tradition, sexual difference that blurs the demarcation between male and female is stigmatized. Until recently, this problem was only slightly familiar to associated medical specialties. We report a case of a patient with 46, XY and point out the dilemma associated with social stigma.

2. CASE PRESENTATION

A 20-year-old Muslim woman who presented to the gynecology outpatient department with primary amenorrhea and delayed pubertal

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Peer review under responsibility of the Dr. Sulaiman Al Habib Medical Services Group Company

features (i.e., poor breast development and scanty pubic hairs) was diagnosed as 46, XY typical male genotype. Result of the systemic examination was unremarkable, whereas genitourinary examination showed scanty pubic hair and blind vaginal pouch. Ultrasound showed remnant testes, whereas uterus, ovary, and fallopian tubes were absent (not visualized). The hormonal assay showed normal luteinizing hormone and follicle-stimulating hormone with low levels of testosterone. Results of the rest of the investigations were unremarkable. The parents and the patient were given a choice of whether she would like to be a male or a female. As the patient grew up as a female for the past couple of years, the patient eventually decided to be female by choice. She underwent surgery; remnant testes were removed, and bilateral breast augmentation and clitoroplasty were performed. As she had presented with a female phenotype at birth, the patient had been raised as a female and developed a consistently female sex identity. The hypothesized cause of the DSD in this patient was complete androgen insensitivity syndrome. She was started on hormone replacement therapy (estrogen in order to maintain her secondary sexual characteristics and bone and cardiovascular health, and to promote general well-being). Moreover, she was advised to undergo follow-up with an endocrinologist.

3. DISCUSSION

Disorders of sex development is congenital abnormality of chromosomal, gonadal, or anatomical sex as proposed by management of intersex disorder and considered a culturally and socially challenging condition; many patients and parents suffer from emotional problems and experience social stigmatization. In many communities, sexual ambiguity and infertility raise disgust, shame, and stigmatization. To avoid such reactions, those with this condition often hide this fact from other people [7,8].

The outer genitalia look female in this case. The birth of a child with sexual uncertainty raises parental feelings of disgrace and guilt. The dilemma for the patients is that they are made to feel completely lonely, and unacceptable [9]. In Pakistan, the situation is further worsened because of illiteracy, which has led to the rejection of intersex people—they are denied education as well as access to healthcare facility. Ignorance of this issue in Pakistani society as well as ignorance in the medical field has led to patients going untreated and living lives of uncontrollable misery that could often leads to internal conflicts and suicide. When their physical structure does not allow them to take part in social life or daily life activities, it often leads to feelings of loneliness, helplessness, and depression [7].

Early diagnosis and treatment is key when aiming to restore normality (physical, social, and psychological) as soon as possible with a multidisciplinary approach including a team consisting trained pediatric surgeons, pediatric endocrinologist, or neonatologists or urologist with their skillful staff along with the help of geneticists, psychologists, ethicists, biochemists, and gynecologists [10,11]. It will not only help in the identification of this rare condition but will also help in the surgical adjustment of the external genitalia, timely gonadectomy, orchidopexy, etc. Basically, the ambiguous genitalia management involves four important steps: precise diagnosis, sex assignment, medical and surgical treatment, and exchange of medical information with parents and patients [12]. History taking with our patient was conducted very carefully with the help of leading questions. Because the patient had presented late to the doctor, the questions related to newborn history may lack specificity and clarity. Discussion of the patient's history with the parents of these patients is always challenging and may lead to agitation and embarrassment.

The attempt to address sex-related issues requires best clinical, biochemical, and genetic tests to ascertain the patient's structural and functional puberty. This whole process requires active participation from the patient and the parents [6]. In this case, the parents and patient were given a choice as to whether the patient would like to be male or female. As the patient grew up as female for the past couple of years, the patient eventually made the choice to be female. The parents have both the moral and legal responsibility and a right to protect the life of their child [13]. Henceforth, the preferences of the parents in regard to sex assignment and management (medically or surgically) of their child are one of the most important factors in the decision-making process. Parents are more familiar with the social and cultural environment in which the child would be raised and how the same influences sex assignment [14]. It is important to assign the sex early so that the child can be raised with a sexual identity. In most developing countries, there is a poor level of awareness and many children with ambiguous genitalia are either missed at birth or present late, which can lead to challenges in sex reassignment [10]. Sex assignment is a difficult task to undergo, both for the individual and the family. In an already polarized society, someone with a third sex is unacceptable for the family and society. The individual is often forced to leave his/her family and community. Intolerance for sex nonconformity is the most critical point in the marginalization of the third sex [15]. In 2018, the Pakistani Parliament introduced a bill that guarantees the rights of the third sex, but laws can only go so far with a community that is so marginalized both economically and religiously [16]. Pakistan, where relationships are so often public property, sex assignment is considered a safeguard.

The topic of ambiguous genitalia has not received due attention from researchers in Pakistan. The fact is that there is minor discussion about external genitalia because of conventional opinions, as well as traditional attitudes of Pakistani people concerning sex and reproductive health privileges. This important matter has been ignored for many years. The important point in the care and social integration of these patients is the much-needed psychological support from family members. Therefore, cultural limitations concerning DSD also obstruct the management process. The clinician, psychologists, and religious leaders can play a vital role in this perspective. The vast research and social awareness campaigns are also an important factor in documenting facts and informing the society about the ambiguous darkness of the taboo associated with intersex disorders. Although diagnosis and treatment are paramount, there is also a need to combat stigmatization associated with this condition, which is widely misunderstood, and the key to this goal is public awareness.

CONFLICTS OF INTEREST

The authors declare they have no conflicts of interest.

AUTHORS' CONTRIBUTION

ZH contributed to acquisition of data. IA, ZH and FK contributed to the conception, write-up and organization of the manuscript. OU and WAK contributed to the critical revision. All authors read and approved the final version of the paper.

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