UIJ UroToday International Journal®

Two Cases of Adult Disorders of Sexual Differentiation Presenting as Hematometra and Adenexal Masses

Adittya K Sharma, Chandrashekhar S Ratkal, Girish Nelivigi, Venkatesh GK

Submitted January 30, 2012 - Accepted for Publication February 21, 2013

ABSTRACT

Disorders of sex differentiation (DSDs) are defined as congenital conditions associated with atypical development of chromosomal, gonadal, or anatomical sex. It presents as a medical emergency at the time of birth, requiring correct gender assignment. In developing countries, adult presentation with incorrect gender assignment is not uncommon. The aim of this paper is to report 2 rare presentations of DSD in adulthood. The first patient presented with hypospadias and a non-palpable right testis while the second patient presented with cyclical abdominal pain and abnormal external genitalia. Both patients were 46,XX on karyotyping, and the computed tomography (CT) of the abdomen showed female internal genitalia with hematometra and gonadal cysts. The delayed adult presentation of DSD, with incorrect gender assignment, is a not a rare occurrence in the developing world. It is a challenging situation, and most of the time management has to be based on psychological and social factors rather than pure scientific principles.

INTRODUCTION

Disorders of sex differentiation (DSDs) are defined as congenital conditions associated with atypical development of chromosomal, gonadal, or anatomical sex [1]. To the best of our knowledge, we are reporting, for the first time, 2 rare cases of adult presentation of DSD as unsuspected hematometra and adenexal cysts.

CASE REPORT

Case 1

A 28-year-old unmarried male presented with a complaint of cyclical suprapubic pain every 2 to 3 months over the past 6 months and abnormal external genitalia since birth. The patient had a history of erections of the phallus upon stimulation without ejaculation. The physical examination revealed a masculine built phenotypic male with a short penis and penoscrotal hypospadias (Figure 1). The scrotum appeared well formed with a small left testis and an absent right testis, which was non-palpable anywhere. The abdominal examination was unremarkable. The contrast-enhanced computed tomography (CT) scan showed a hematometra with a heterogeneously enhancing right adenexal mass and a large cyst (Figure 1). The uterus appeared to be communicating with the posterior urethra with a fistulous tract. His hormonal profile was normal for a male with serum testosterone on the low/normal side. An evaluation of congenital adrenal hyperplasia was negative. After karyotyping, the diagnosis of 46,XX DSD was made. The patient underwent a laparoscopic hysterectomy with an excision of the right adenexal mass (Figure 1). The histopathology confirmed benign right-sided ovotestes with a cystic component, along with hematometra and endometritis. Since the patient wanted to continue with his male phenotype, he was counseled regarding impotence and infertility. Later, the patient was planned for the correction of his hypospadias.

Case 2

KEYWORDS: Hermaphroditism, hematometra, DSD

CORRESPONDENCE: Adittya K. Sharma, MCh, Institute of Nephrourology, Victoria Hospital Campus, Bangalore, Karnataka, India (dradityaonline@gmail.com, dradityaks@gmail.com)

CITATION: *UroToday Int J*. 2013 April;6(2):art 25. http://dx.doi.org/10.3834/uij.1944-5784.2013.04.12

Figure 1. a) Adult male phenotype; b) A short penis with a small left testis and a non-palpable right testis; c) Female internal genitalia; d) Gross specimen of the uterus with a right adenexal mass.

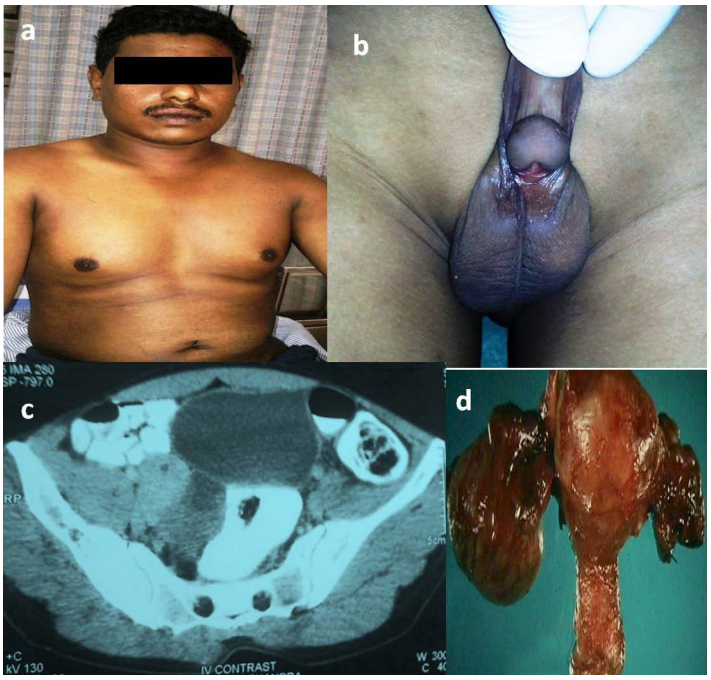
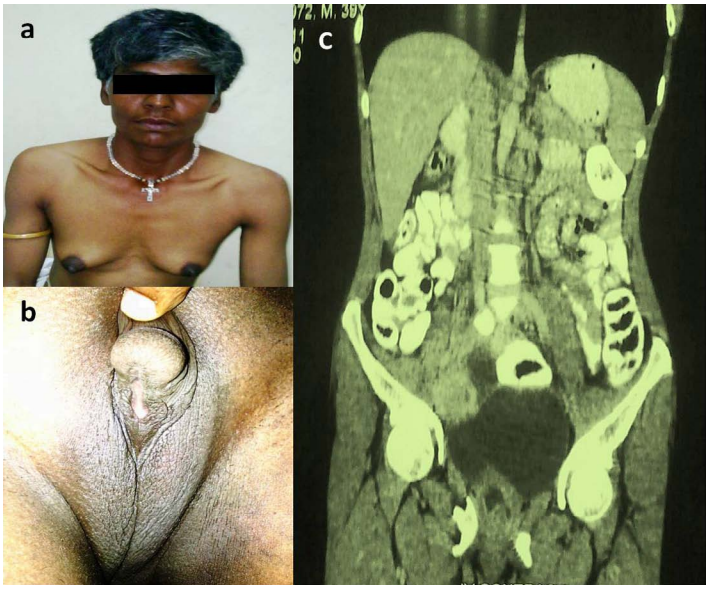


Figure 2. a) Adult female phenotype; b) Ambiguous external genitalia; c) The uterus with a hematometra and an adenexal mass.



A 24-year-old mentally challenged male presented with complaints of abnormal external genitalia. An examination showed a thinly built phenotypic female with well developed breasts, hirsute facies, and female-pattern distribution of hair (Figure 2). External genitalia revealed a small phallus with a dorsally hooded prepuce with penoscrotal hypospadias. The scrotum was bifid and hypoplastic with absent testicles and no vaginal opening (Figure 2). The patient was brought up as a male child with no prior medical consultation for his condition. A radiological evaluation revealed the presence of a hematoma in the uterus and a thickened endometrium with a cyst in the left adenexa (Figure 2). A biochemical evaluation showed serum FSH, LH, and progesterone levels were normal while his serum testosterone levels were high for a female. Karyotyping confirmed the diagnosis as 46,XX DSD. The patient was brought in with an expectation of converting the appearance of his external genitalia to that of a male. After counseling regarding the prospective usefulness of his extensive surgical correction, relatives chose against any intervention.

DISCUSSION

The word intersex has conventionally been used to refer to the appearance of the external genitalia but it is now considered inappropriate [2]. The Chicago consensus in 2005 established revised nomenclature and treatment recommendations in individuals with the newly defined term "disorders of sex differentiation" (DSDs), replacing terms such as intersex, hermaphroditism, and pseudohermaphroditism [1,3,4].

Criteria of physical findings suggestive of DSD include: 1) overt genital ambiguity (e.g., cloacal extrophy); 2) apparent female genitalia with an enlarged clitoris and posterior labial fusion (e.g., CAH); 3) apparent male genitalia with bilateral undescended testes, hypospadias, or micropenis; and 4) discordance between genital appearance and a prenatal karyotype [1]. Most DSDs are diagnosed in the neonatal period. Later presentations in older children often include: 1) previously unrecognized genital ambiguity, 2) inguinal hernia in a girl (e.g., complete androgen insensitivity), 3) delayed or incomplete puberty, 4) primary amenorrhea or virilization in a girl, 5) breast development in a boy, and 6) gross or cyclic hematuria in a boy [1]. Given the spectrum of findings and diagnoses, no specific single protocol could be used in the evaluation of DSD patients.

Adult presentation is significantly challenging as in mentioned cases because of incorrect assignment of sex at the time of birth. Unawareness about the need of seeking medical help is one reason and desire of a male child, hence tagging anything ambiguous accordingly, which affected our second case. Not surprisingly, in both cases correction to male phenotypes was desired, which was justifiable in the first patient who had a

male psychological orientation but not advisable in the second patient who lacked sexual understanding.

Initial gender uncertainty is distressful news for families. Influencing factors to consider when discussing gender assignment include diagnosis, genital appearance, fertility potential, therapeutic/surgical options, and familial views or circumstances relating to cultural biases. The general recommendations are to raise infants with 46,XX CAH or 46,XY CAIS as females, whereas infants diagnosed with 5-alpha reductase deficiency or 17-beta hydroxysteroid dehydrogenase 3 deficiencies, a male assignment should be considered [5].

But in adult presentations, psychological orientation plays the pivotal role in decision-making. The surgical correction technique and the timing of the operation need to be individualized according to medical conditions, experience of the surgeon, and the complexity of each case. The general trend is toward early reconstruction with subsequent early and long-term management of the patient.

CONCLUSION

Current recommendations emphasize sensitive, supportive interactions with families, and full disclosures of the risks, benefits, and potential outcomes of intervention to allow them to participate as fully as possible in decision-making and in the continued care of their child.

REFERENCES

1. Lee, P. A., C. P. Houk, et al. (2006). "Consensus statement on management of intersex disorders. International Consensus Conference on Intersex." *Pediatrics* 118(2): e488-500. PubMed | CrossRef

2. Conn, J., L. Gillam, et al. (2005). "Revealing the diagnosis of androgen insensitivity syndrome in adulthood." *BMJ* 331(7517): 628-630. PubMed | CrossRef

3. Hughes, I. A., C. Houk, et al. (2006). "Consensus statement on management of intersex disorders." *Arch Dis Child* 91(7): 554-563. PubMed | CrossRef

4. Houk, C. P., I. A. Hughes, et al. (2006). "Summary of consensus statement on intersex disorders and their management. International Intersex Consensus Conference." *Pediatrics* 118(2): 753-757. PubMed | CrossRef

5. Mendonca, B. B., M. Inacio, et al. (2003). "Male pseudohermaphroditism due to 5 alpha-reductase 2 deficiency: outcome of a Brazilian cohort." *Endocrinologist* 13: 201-204. CrossRef