CASE REPORT

COMPLETE TESTICULAR FEMINIZATION A CASE REPORT AND REVIEW OF THE LITERATURE

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ABSTRACT
Complete Testicular Feminization (CTF) is an androgen receptor function disorder. The diagnosis may be made in the postpubertal period on the basis of clinical and hormonal findings accompanying amenorrhea, a karyotype analysis of 46 XY with a female phenotype and absence of pubic hair or inguinal hernias containing testes. In patients with CTF, bilateral undescended testes carry a high risk for malignancy and the risk of malignant transformation of the gonads in adult life is well documented. Testicular feminization often can lead to problems in relations or marriages, and even sometimes result in social disasters. Physicians must be very careful in selecting correct words in order to explain the situation to the patient and family, especially in conservative societies. Here, we present a case of a 30-year old woman with primary amenorrhea who was diagnosed as CTF and underwent bilateral orchiectomy.

Keywords: Complete testicular feminization, Androgen receptor function disorders, Pseudohermaphroditism

INTRODUCTION
Disorders of androgen receptor function represent the most common detactable cause of male pseudohermaphroditism. These patients characteristically have a 46 XY karyotype and present with a spectrum of phenotypic abnormalities. CTF is the most severe form of androgen receptor function disorders. This condition has an incidence of 1 in 20000 to 1 in 60000 males and it is transmitted as an X-linked trait. CTF syndrome should be suspected in a girl in the presence of inguinal hernia without genital ambiguity at birth. During embryogenesis, dihydrotestosteron (DHT) stimulates male external genitalia differentiation. Significantly decreased or abnormal DHT receptors are responsible from the pathology in CTF syndrome. In utero resistance to testosterone action prevents stabilization of the Wolffian ducts. However, because the fetal testes secrete MIS (Müllerian Inhibiting Substance), internal genitalia are absent with the exception of the testes, which may be found in the labia, inguinal canal or the abdomen. Endocrine evaluation in the newborn period demonstrates normal levels of testosterone, DHT and gonadotropins. At puberty, gonadotropin levels rise, leading to increased...
levels of plasma estradiol, which results in feminization, including breast development. The diagnosis of CTF may readily be made in the postpubertal patient on the basis of clinical and hormonal findings of amenorrhea, absence of pubic hair, or inguinal hernias containing testes. It is confirmed by a 46 XY karyotype and a normal male androgen and gonadotropin profile. Pelvic ultrason examination confirms the absence of Mullerian tissue and a vaginal examination confirms a blind ending vagina without a cervix 4.

**CASE REPORT**

A 30-year-old married woman with a normal female phenotype admitted to our outpatient clinic with a history of infertility and primary amenorrhea. Breast development was normal, pubic and axillary hair was scarce. Physical examination revealed a blind-ending 5-6 cm long vagina whereas cervix and uterus were nonpalpable. In digital rectal examination, a small prostate was palpable. Hormonal analysis showed a total testosterone of 10.7 ng/ml, estradiol: 65.7 pg/dl, prolactin: 11.7 ng/ml, FSH: 21.5 mIU/ml, LH: 16.4 mIU/ml. Karyotype analysis was reported to be 46 XY.

Abdominal ultrasonographic examination showed bilateral undescended testes of 3-3.5 ml volume in the inguinal canals proximal to the external inguinal ring without hernia. No uterus or ovaries were detected by ultrasound. Abdominopelvic CT revealed a normal prostate gland and two seminal vesicles posterior to the urinary bladder. After the diagnosis of CTF, the patient was informed about the situation. With the consent of the patient, bilateral inguinal orchiectomy was performed and estrogen replacement therapy (ERT) was started 2 weeks after the operation. She is still on estrogen replacement therapy. Meanwhile, as a social and cultural consequence, the couple was divorced immediately after getting the karyotype result, because of the strong opposition of the husband’s family to the continuation of the marriage.

**DISCUSSION**

In CTF, bilateral undescended testes carry a high risk for malignancy. The risk of malignant transformation of the gonads in adult life is well documented 5,6. Incidental seminoma has also been reported by Sahai et al in a patient with CTF who underwent bilateral orchiectomy 7. The best time for orchiectomy to those patients is soon after completion of puberty. The point here is that estradiol produced from testes is necessary for a normal phenotypic female pubertal development. After orchiectomy, estrogen replacement therapy should be started. However, if bilateral orchiectomy is performed before puberty as a result of testicular trauma, testicular torsion, or severe pain, estrogen replacement therapy will be necessary.

Since nearly half of the patients had a proved family history of CTF, it appears that more attention should be given to genetic counselling for the families of CTF patients. Unfortunately, it is to note that a proved family history is the mode of presentation in only 14% of cases 8. A very important step in the treatment of CTF patients is sex determination. Most patients are satisfied with their sexual functioning and are happy to be female. Wisniewski, et al. reported that 78% of patients with CTF syndrome were satisfied with their genitalia in terms of sexual function. They also reported that 71% of the patients had orgasm and were satisfied with their overall sexual function 9. The large majority of patients in this study reported a normal female heterosexual orientation in terms of sexual attraction. Additionally, patients were satisfied with their physical appearances. As a result, non of the patients had a desire to change their sex 9. However, as in our case, testicular feminization often can lead to problems in relations or marriages, and even sometimes result in social disasters. Physicians must be very careful in selecting correct words in order to explain the situation to the patient and family, especially in conservative and under-educated societies. For couples who want to continue the relation and want to have children child adoption should be advised.

**REFERENCES**

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