TRUE HERMAPHRODITE - A CASE REPORT

C H Yip, R Pathmanathan

ABSTRACT

A case report of a male true hermaphrodite with 46XX/46XY karyotype is presented. He was first diagnosed at the age of 9 years when he presented with hypospadias and a left undescended testis. He was lost to follow-up until he presented at the age of 23 years with bilateral gynaecomastia. A hormonal profile showed a low testosterone level, while a seminal assay showed very few sperms. However he claimed to be sexually active. A year later, after he got married, he began to complain of impotence. A review of the condition is presented.

Keywords: *true hermaphrodite, hypospadias, bilateral gynaecomastia*

SINGAPORE MED J 1996; Vol 37: 117-118

INTRODUCTION

The term "hermaphrodite" is derived from Hermaphroditus, who, in Greek mythology, was the son of Hermes and Aphrodite. He united with the nymph of the Fountain of Salmacis, the two forming one person with characteristics of both sexes.

A true hermaphrodite is an individual whose gonads contain ovarian tissue with follicles, and testicular tissue with spermatozoa or seminiferous tubules. This condition has to be differentiated from mixed gonadal dysgenesis where there is a unilateral testis and contralateral dysgenetic "streak" gonad[1]. True hermaphrodites are uncommon. There are about 750 cases reported in the literature since 1899. The expectation of a satisfactory sexual life for a true hermaphrodite is poor, because of the immature development of the gonads. We present here a male true hermaphrodite.

CASE REPORT

MM is an Indian male child who was first seen at the age of 9 years, when he sought treatment for penoscrotal hypospadias and a left undescended testis. Reconstruction of the hypospadias was done, and exploration of the left inguinal canal carried out. The left testis was found to be intraperitoneal and rudimentary. No uterus was found. The left gonad was removed and found to contain both ovarian and testicular tissue. The right testis was biopsied and showed only testicular tissue. Chromosome studies showed a 46XX/46XY karyotype. He recovered uneventfully and was discharged. He defaulted any further follow-up, and was next seen at the age of 23 with bilateral gynaecomastia. He reported normal sexual activity with his girlfriend and was able to achieve erection, penetration and ejaculation. However the amount of semen was small.

On examination, he was a masculine looking young man with a moustache. However he had a female body shape, with well-developed breasts and female distribution of pubic hair. His penis was 3.5 cm long, and his right testis was 10 cc volume measured with an orchidometer. The left testis was absent (Fig 1).

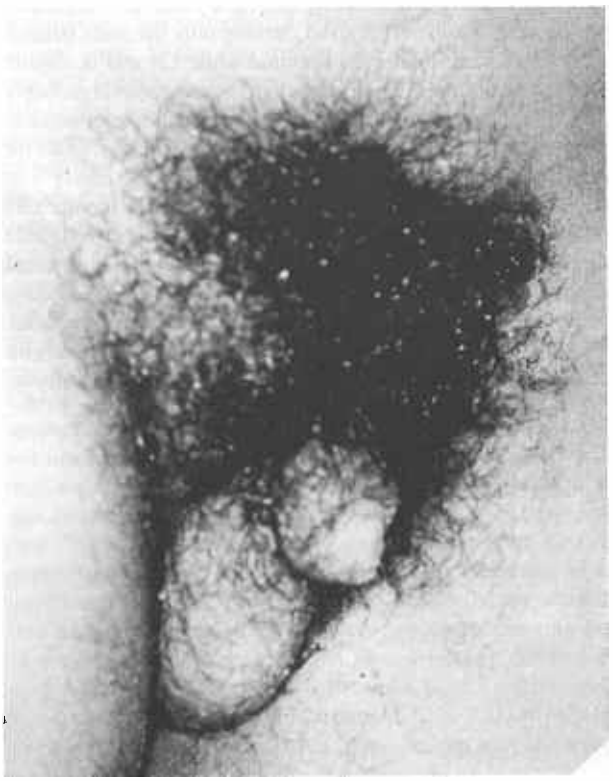
Hormonal profile showed a progesterone level of less than 1 nmol/L, oestradiol of 117 pmol/L and luteinising hormone of 13 IU/L. These were all within the normal values for an adult male. However the follicular stimulating hormone was elevated (27 IU/L) and testosterone level was low (5.9 nmol/L). A sperm analysis showed a volume of 2.3 ml, pH of 6.5, and only a few sperms were seen. A bilateral subcutaneous mastectomy was done.

He was started on monthly intramuscular testosterone enanthate 250mg. He married soon after the surgery, but within a year, complained of decreasing potency, with inability to sustain an erection, despite testosterone therapy.

DISCUSSION

True hermaphrodite is the most uncommon variant of intersexuality in the human being. The majority of them have

Fig 1 – Genitalia showing a small penis, absent left testis and female distribution of hair



Department of Surgery
Faculty of Medicine
University of Malaya
59100 Kuala Lumpur
Malaysia

C H Yip, MBBS, FRCS (Glasg)
Associate Professor

Department of Pathology
University of Malaya

R Pathmanathan, MBBS, MRCPath, FRCPA
Professor

Correspondence to: Dr C H Yip

the 46XX karyotype[2]. The 46XX/46XY mosaicism, which is present in our patient, is the next commonest type. There have also been reports of 46XY, and 45XO true hermaphrodite. The commonest gonadal type is an ovary-testis[3]. The ovotestis-testes type, which this patient has, occurs in about 10% of true hermaphrodites.

The aetiology and pathogenesis of true hermaphrodites is unknown. The rare occurrence of true hermaphrodites in siblings[4] and the fact that true hermaphrodites is the commonest form of intersex states in South African Blacks[2,3] indicate that there might be a genetic or epidemiological factor involved.

The commonest presentation is an abnormal external genitalia[2,3] and this prompts parents to seek medical advice early on in life. The next commonest presenting symptom is during puberty, when a male suddenly starts developing breasts. A rare case of a male true hermaphrodite presenting as a male "transsexual" has also been reported[5]. Other presenting symptoms are cryptorchidism, haematuria, amenorrhea, lower abdominal pain, inguinal hernia, and hypospadias. Since 75% of true hermaphrodites are raised as males, breast development is a significant presenting symptom.

In the management of true hermaphroditism, the first priority is to establish beyond any doubt the gender of the patient[6]. In the patient below the age of 1 year, the sex of rearing is determined by the functional potential of the external genitalia. Female-directed plastic surgery is much more successful than attempts to construct male genitalia[7]. However this patient was diagnosed at 9 years of age, and psychological assessment at that time showed that he was very strongly male-oriented, and sex reassignment at that age would pose serious psychiatric problems. Hence, in patients with peno-scrotal hypospadias and cryptorchidism, the possibility of true hermaphroditism should be considered, and gonadal exploration and biopsy carried out preferably before the age of 2 years. In the patient with a male gender role, care should be taken to remove all ovarian tissue as well as female organs. Supplementary testosterone will be required during puberty if testosterone levels are low.

There are very few data available on FSH, LH, testosterone, and oestradiol values in the plasma of patients with true hermaphroditism during the reproductive years. In this patient, the ovotestis has been removed, leaving only the testis behind. FSH levels were higher than normal while LH and oestradiol levels were normal. This suggests that ovarian tissue is probably absent, although it is difficult to explain the gynaecomastia in this patient. The testosterone level was half the normal value for males, indicating the immature development of the testis.

The expectation of a satisfactory sexual life for the true hermaphrodite is poor[2,8]. There have been a few reports of female true hermaphrodites who have married, became pregnant and delivered normal children[9]. However reports of sexually active male hermaphrodites are rare[10]. Fertility is greatly reduced. There was one reported case of a male hermaphrodite with cryptorchidism, who had a documented normal sperm analysis, and fathered two children[11]. In this patient he initially reported satisfactory sexual intercourse but he later developed impotence, probably due to the immature development of the remaining gonad. The sperm count showed that he was unlikely to father any children even if he was potent.

True hermaphroditism is rarely associated with gonadal tumours, unlike in mixed gonadal dysgenesis, where the presence of a dysgenetic gonad predisposes to gonadal malignancy. However a few cases of malignancies like dysgerminoma and gonadoblastoma have been reported in the true hermaphrodite[12-14]. Hence this patient will require close follow-up to diagnose any malignancy arising in his remaining testis. Since the incidence of gonadal malignancy is low, estimated at 4.6%[2], prophylactic removal of his remaining testis is probably not justified.

References

1. Davidoff F, Federman DD. Mixed gonadal dysgenesis. Pediatrics 1973;52:725-42.
2. Krob G, Braun A, Kuhnle U. True hermaphroditism: geographical distribution, clinical findings, chromosomes and gonadal histology. Eur J Pediatr 1994;153:2-7.
3. van Niekerk WA. True hermaphroditism. An analytical review with a report of 3 new cases. Am J Obstet Gynecol 1976;126:890-907.
4. Gallegos AJ, Guizar E, Armendares S, Cortes-Gallegos V, Cervantes C, Bedolla N, et al. Familial true hermaphroditism in 3 siblings: Plasma hormonal profile and *in vitro* biosynthesis in gonadal structures. J Clin Endocrinol Metab 1976;42:653-60.
5. Kok LP, Tsoi WF. A case of a hermaphrodite presenting as a transsexual. Ann Acad Med Singapore 1982;11:297-9.
6. Tay BL. Intersex and genital ambiguity. Singapore Med J 1990;31:255-8.
7. Oesterling JE, Gearhart JP, Jeffs RD. A useful approach to early reconstructive surgery of the child with ambiguous genitalia. J Urol 1987;138:1079-82.
8. Aaronson IA. True hermaphroditism. A review of 41 cases with observations on testicular histology and function. Br J Urol 1985;57:774-9.
9. Narita O, Manba S, Nakanishi T, Ishizuka N. Pregnancy and childbirth in a true hermaphrodite. Obstet Gynecol 1975;45:593-5.
10. Mc Govern JH, Marshall VF. Three cases of true hermaphroditism. J Urol 1962;88:680-90.
11. Shannon R, Nicolaides NJ. True hermaphroditism with oogenesis and spermatogenesis. Aust NZ J Obstet Gynecol 1973;13:184-7.
12. McDonough PG, Rogers Byrd J, Phung TT, Otken L. Gonadoblastoma in a true hermaphrodite with 46XX karyotype. Obstet Gynecol 1976;47:355-8.
13. Szokoi M, Kondrai G, Papp Z. Gonadal malignancy and 46XY karyotype in a true hermaphrodite. Obstet Gynecol 1977;49:358-60.
14. Park IJ, Pyeatte JC, Jones HW, Woodruff JD. Gonadoblastoma in a true hermaphrodite with 46XY genotype. Obstet Gynecol 1972;40:466-72.