A Case of True Hermaphroditism Presenting as an Undescended Testicle

R. H. JOHNSON

SUMMARY

A case of true hermaphroditism in a 20-year-old Motswana who was brought up as a male, is reported. He presented with left-sided undescended testicle and inguinal hernia. At operation an ovary, uterus and Fallopian tube were found instead of a left testicle.

Chromosome analysis revealed a karyotype, 46, XX in tissue culture and peripheral blood. Epithelial nucleii in buccal smears were chromatin-positive, indicating the presence of 2 X chromosomes.

S. Afr. Med. J., **48**, 1540 (1974).

True hermaphroditism is the commonest intersex state in the Black population of South Africa,[1] with an estimated incidence of 1/10 000 live births.[2] The case reported here is believed to be the first reported in a Motswana.

CASE REPORT

A 20-year-old Motswana, brought up as a male, was referred because of an undescended left testicle. No testicle could be found in the scrotum, perineum or inguinal region. A left inguinal hernia of the indirect type was noted. He also had a small right testicle, penile hypospadias and mild bilateral gynaecomastia. There was also an absence of facial hair — not uncommon in young Motswana males.

Exploration of the groin demonstrated a small ovary, Fallopian tube and a structure resembling a small uterus, instead of a left testicle. Because the patient was by appearance and upbringing a male, these were removed and the inguinal hernia repaired by Bassini's method. The patient made an uneventful recovery.

The organs removed were sent for histological examination the Royal Army Medical College, London, as there was no pathologist in Botswana. The sections of the uterus revealed a normal myometrium, and the endometrial glands had stratified epithelium, but were inactive. The Fallopian tube was normal. The structure resembling an ovary did consist of ovarian stroma, and contained normal primary follicles and follicular cysts. There was no testicular differentiation.

Princess Marina Hospital, Gaborone, Botswana
R. H. JOHNSON, F.R.C.S., *Surgeon* (Present address: 45 Henry Drive, Leigh-on-Sea, Essex, England)

Date received: 28 March 1974.

After consultation with Dr E. Wilton of the Cytogenetics Unit of the South African Institute for Medical Research in Johannesburg, a specimen of peripheral blood, a skin biopsy specimen and a buccal smear were sent for karyotype analysis. This showed a model number karyotype 46, XX, with no gross morphological abnormality on light microscopy. The buccal smear showed epithelial nuclei to be chromatin-positive, indicating the presence of 2 X chromosomes. This was consistent with the presence of ovarian tissue in the patient and the diagnosis of true hermaphroditism.

DISCUSSION

As mentioned, true hermaphroditism is the commonest cause of intersex problems in the Black population of South Africa, with an incidence estimated at 1/10 000 live births. Wilton,[1] in an analysis of 238 patients of all races with anomalous sexual development, from major hospitals on the Witwatersrand, found 46 patients to be true hermaphrodites. All were Black, although Grace and Edge[2] have since reported a case of true hermaphroditism in a White infant. On the other hand, Wilton found Klinefelter's syndrome to be uncommon among Blacks and more common among Whites.

Mosaicism has not been entirely excluded in this patient, since no organ cultures were done either from the specimen or the right testicle.

In Klinefelter's syndrome, spermatogenesis has been shown to be affected adversely in patients with a 47,XXY karyotype, compared with patients who have 46,XY/47,XXY mosaicism. The presence of the extra chromosome is said to be damaging to the testes.[3] The probability is that this patient will therefore be of very low fertility at best, although not impotent, since he does have penile erections.

Pyschological complications can occur if the sex of rearing is at variance with the psychological sex, but this patient was in appearance a male, had been brought up as such, and therefore ought to be well adjusted.

I should like to thank Dr E. Wilton of the Cytogenetics Unit of the South African Institute for Medical Research, Johannesburg for the karyotype analysis, and Major R. L. Warkel of the Histopathology Registry, Royal Army Medical College, London, for the histological report.

REFERENCES

1. Wilton, E. (1969): S. Afr. J. Surg., **7**, 45.
2. Grace, H. J. and Edge, W. E. B. (1973): S. Afr. Med. J., **47**, 1553.
3. Gordon-Donald, L., Krmpotic, Eva, Thomas, W., Gendy-Hortense, M. and Paulsen, C. A. (1972): Arch. Intern. Med., **130**, 726.