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Two Cases of Transsexualism with Gonadal Dysgenesia

By B. IONESCU, C. MAXIMILIAN and A. BUCUR

Most psychiatrists who have made a study of abnormal psychosexual development accept Money's postulate as to the manner in which heredity and education determine sexual development. This assumes that genetical and hormonal factors influence psychosexual development only through bodily shape, while sexual behaviour is learned like the native language. Nevertheless, Money himself raises the question as to whether patterns of sexual play, activity and distractibility in the two sexes are not elements of a strong genetical determinism.

At birth, sexual assignment (as male or female) and rearing are determined by the external morphology of the genitals, and education usually follows this assignment. A sense of sexual identity (masculinity or femininity) is then built up gradually in accordance with bodily shape and gender role, developed by education and the personal experience of the individual

There are many cases of psychological disorders of sex that cannot be explained by assuming that psychosexual development is decided either by external genital form or by education alone. A similar conclusion was reached by Van Wyk and Grumbach (1968) in a study of the 'sex of rearing'.

Somatic sexualization is thought to be due to the steady influence of the sexual chromosomes acting through the gonads. It seems that psychological sexualization is also determined by genetical factors, and this is illustrated by the following case histories of psychosexual problems.

Case 1.

F.N. A 26-year-old bricklayer. At birth he was declared male in accordance with his external genitalia. Birth, growth and development were apparently normal, and at 15 or 16 pubic hair, penis and scrotum developed and his voice broke.

On examination. He was 162 cm. tall and weighed 72 kg. Body-form was gynaecoid, with much adipose tissue in the lower abdomen, thighs and buttocks. The mandible was prominent, and hands and feet were large. As to secondary sexual characteristics, there was a localized excess of adipose tissue in the breasts and the nipples were small and pale. Axillary hair was scanty. Pubic hair was well developed but triangular in outline. The skin was smooth.

The external genitals were masculine. Penis, scrotal skin and prostate were normal and cremasteric reflex present. However, the scrotal contents were a small, soft, ill-defined structure on the left and a gelatinous mass the size of an olive on the right hand side.

Investigation. A buccal smear was chromatin negative, and chromosomal analysis of leucocytes cultured from peripheral blood demonstrated a normal male karyotype 46 XY.

Urinary 17 ketosteroids 4.84 mgm. per 24 hours (normal 10 to 25 mgm.).

Twenty-four hours urinary gonadotropins 20 mouse uterine units.

Twenty-four urinary total oestrogens 23.5. mgm.

Urinary pregnane — 3 alpha, 17 alpha, 20 alpha — triol of 0.33 mgm. per 24 hours.

17-ketosteroids and oestrogens excretion was not modified by stimulation with gonadotrophins or supression of the adrenal cortex.

Laparotomy. No reproductive structures in the pelvis.

Biopsy. Bilateral incisional testicular biopsies revealed on the left side, fibrous mesenchymal tissue; and, on the right, a hypoplastic male gonad of tubules with a thin basement membrane and seminiferous epithelium (consisting mainly of Sertoli cells and some spermatogonia). The interstitial tissue was made up of non-specific elements.

Sexual behaviour. Although declared to be a boy and brought up as one, the subject was dressed at home as a girl and told to help his mother with the housework. He felt sure throughout his childhood that he was a girl, and was shocked when required to dress as a boy on going to school. From the age of 7 to 12 at school he was regarded sometimes as a boy and at others as a girl. He preferred to play with girls and liked needlework. From 14 to 24 in residential establishments he shared a bedroom with men until his feminine behaviour annoyed his room mates. Subsequently, he was allowed to wear women's clothes and reside with females.

While an in-patient at the Institute of Endocrinology his behaviour was noted to be feminine, and he made frequent approaches to men. He felt strongly attracted by men and had no sexual interest in women. He was greatly troubled by his predicament. He abhorred the idea of the male sexual role, but also that of passive sodomy. He felt at ease dressed as a woman and very uncomfortable dressed as a man.

Case 2.

R.G. A 31-year-old electrical engineer had been declared female at birth in accordance with her external genitalia.

Birth, growth and development were normal. Breasts began to develop at the age of 15. Menstruation began at 17 or 18, and ceased at 28. Immediately after cessation of the menses, the body became hairy, especially on the upper lip, chin and linea alba, while facial acne appeared.

On examination. The body form was android and juvenile, resembling that of a boy of 17. Body hair was moderately developed, with triangular distribution of pubic hair and hairiness on the linea alba. Hair was abundant on the face, between the breasts and around the nipples. The voice was baritone. Breasts were well developed with normally pigmented nipples. External genitalia were female, but with a hypertrophic clitoris (about 3 cm. on the dorsal aspect). The vagina was normal and 10 cm. long.

Investigation. A buccal smear was chromatin positive. Chromosomal analysis of leucocytes cultured from peripheral blood demonstrated a normal female karyotype 46 XX. Hormonal levels following stimulation with gonadotrophin and suppression of adrenal cortex are shown in Table I.

Laparotomy. This revealed a microfibromatous median uterus and normal Fallopian tubes. The left ovary was normal in size and had a white mother-of-pearl coloured albuginea. The right ovary had the same appearance but for a cyst the size of a hen's egg. Bilateral incisional ovarian biopsies revealed a hyperplastic stroma with normal germinal tissue and follicles in various degrees of maturity but none at the pre-ovulation stage. The persisting follicles showed a granulo-thecal hyperplasia.

Sexual behaviour. From early childhood she considered herself a boy and claimed to be dressed as such. She rejected explanations to the contrary, and was punished for her contrariness. Later, however, she deferred to her parents' requirements and dressed as a female, feeling withdrawn and misunderstood. At the age of 15 she felt erotically attracted by girls. She was deeply depressed by the development of breasts and the menarche, and by an unsuccessful attempt at the male role in coitus. She regarded herself as a 'monster', a 'mockery of nature', hated her body, and at the age of 22 attempted suicide. Subsequently, unable to regard herself as female, she suppressed erotic desires but continued to dress as a man. At the age of 28 when the menses stopped and facial hair grew her conflicts reappeared. She supposed herself to be male after all, and to have evidence that would persuade others accordingly. She appealed to the Institute to support her contention. In hospital, she behaved as a polite, reserved man, accepted socially as such by people of both sexes.

Diagnosis

These cases (the first a man and the second a woman) were diagnosed as transsexualist on the following grounds:

(a) The subjects contested their sexual assignment from early childhood, and maintained their belief contrary to external physical signs or educational coercion.

(b) They could not convince themselves, even when they tried, that their psychological sex corresponded with their physical appearance.

(c) They desired sexual activity appropriate to the sexual identity which they claimed, and denied sexual perversion.

(d) In order to have their desired sexual identity confirmed they were prepared for surgical procedures.

(e) Their attempts to achieve recognition of their desired sexual identity was based more on a wish to be rid of 'an error of nature' than to obtain erotic satisfaction.

The transsexualist considers it natural and self-explanatory that he should wear clothes corresponding to his psychological sex. Doing so brings him peace of mind rather than sexual excitement; he wears his 'travesty' unobtrusively and out of a sense of necessity. By contrast, the transvestite is motivated by a desire for ostentation and masquerade.

The first subject was genetically male, but had only one testis, and that very abnormal. Testicular activity during the embryo-fetal period induced male genital morphology, but male sexualization did not occur, and gynaecoid features and high oestrogen levels were evident. The diagnosis was testicular dysgenesis with transsexualism.

In the second case, a normal female karyotype (46 XX), congruous external genitalia, and prolonged ovarian activity were associated with

Table I

	Case	17—K	DHA	A + E	11—OH	ES—T
Starting point	1	4·840	1·820	2·160	0·860	23·5
	2	2·883	0·048	1·875	0·960	23·6
Inhibition with dexamethason	1	1·422	0·110	1·150	0·162	21·1
	2	1·242	0·030	1·000	0·212	20·1
Dexamethason and prolan	1	1·215	0·050	0·893	0·272	36·2
	2	1·115	0·000	0·868	0·247	38·5

17—K: 17—Ketosteroids; A + E: Androsteron + Etiocolanolon; DHA.: Dehidroepiandrosteron; 11—OHr: 11—Oxicorticosteroids; ES—T: Total Estrogens.

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android body shape, clitoral hypertrophy and granulothecal hyperplasia of the ovaries. The diagnosis was ovarian dysgenesis with transsexualism.

Discussion

Transsexualism has been reported in cases of underdeveloped gonads (Benjamin, 1966), oestrogen-secreting testicular tumours (Stoller *et al.*, 1960), feminizing suprarenal tumours (Routier *et al.*, 1964), and Klinefelter's syndrome (Brown *et al.*, 1964; Davidson, 1966; Klotz *et al.*, 1955; Money, 1967; Pauly, 1965).

Other disorders of psychosexual orientation, such as homosexuality or transvestism, occur more frequently in cases of gonadal dysgenesis than is usually supposed (Dowling *et al.*, 1963; Ionescu *et al.*, 1969; Aubert *et al.*, 1966).

The normal female zygote is 46XX, the normal male 46XY. Ovarian dysgenesis produces a female with incomplete feminine characteristics. The somatic sexualization of the normal male is attributable to the Y chromosome, which promotes development of the testis, which in turn moulds the phenotype against the feminizing tendencies of the X chromosome. In testicular dysgenesis, the testis fails either to induce male development or to suppress the feminizing influence of the X chromosome. These somatic sexualizing processes are well known, but those of psychic sexualization are imperfectly understood.

Important recent studies of the rat indicate that this animal has two hypothalamic centres, one of which is responsible for 'sexual behaviour' and the other for 'hypophyseal function' of a masculine or feminine pattern. The function of these centres seems to be genetically programmed for both sexes according to a feminine pattern (Barraclough *et al.*, 1964; Harris *et al.*, 1964, 1965, 1966; Young *et al.*, 1965). Unless masculinizing factors operate during the perinatal period, the role of these centres remains feminine throughout life. The perinatal operation of masculine hormones induces masculinity through the hypophyseal centres whether the individual is male or female.

Education fosters a pattern of part-functions of psychosexual development. But despite the well-known Harlow (1965) experiment on the learning of sexual play by apes, there is good evidence of intrinsic elements which determine human sexual behaviour (Young *et al.*, 1965; Schlegel *et al.*, 1967; Mizuno *et al.*, 1968). The first post-natal days are crucial in psychic sexualization, deviation in which can be produced at this time by androgens in the female, or lack of them in the male.

Gonadal dysgeneses induce abnormal psychic sexualization only when they distort the perinatal stage of development. Certain cases of psychosexual disorder may be caused by disturbance of the hypothalmic centres during the perinatal stage without any subsequent physical evidence of gonadal dysgenesis.

Sexualization of the individual takes place in linked stages. There are a number of sexual variables and these may be independent of one another. Discordance may occur between stages, among elements of the same stage or in both respects. Sometimes an incongruous element of one stage may remain latent until a later stage when its sexualizing influence becomes manifest.

If the development of psychic sexualization and sexual behaviour are seen as intrinsic parts of the same process, then disturbances of them are to be regarded as states of intersexuality in which concordance between psychic and somatic sex is lacking.

Traditional forms of psychotherapy have been totally unsuccessful in their emphasis upon the conflicts which supposedly underly symptoms of intersexuality in adults. We join those authors (Baker, 1969; Pauly, 1968; Stoller, 1964) who contend that sex-conversion operations are generally successful. Nevertheless the variety of clinical forms, such as transvestism and transsexualism, in which intersexuality is manifest, and the degrees of severity of these conditions, seem to be determined by education and experience.

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B. Ionescu, M.D.
C. Maximilian, M.D.
A. Bucur, M.D.
Institutul de Endocrinologie, Bulevardul Aviatorilor 34, Bucharest, Rumania

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