A Case of Female Intersex

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SUMMARY

A case of female intersex was admitted to the hospital with severe lower abdominal pain. On examination a phallus of 5 × 2,5 cm was found, without a urethra but with a perineal urinary opening. There were two scrotal folds, but no vagina. The patient had the build of a female, with two well-formed breasts.

At operation a reasonably small uterus (5 × 3 × 2 cm), was found with normal Fallopian tube and round ovary on the right side. There was an acute ovarian abscess on the left side.

Diseased tissue was removed and the abscess drained.

The multiple problems and embryological disturbances of the patient and his albino Black wife are discussed with reference to available literature.

S. Afr. Med. J., **48**, 465 (1974).

CASE REPORT

A 29-year-old Black patient, apparently male, was referred to hospital (for the sake of simplicity the male gender is used), complaining of severe lower abdominal pain, radiating down the left thigh.

The pain had started 4 days previously being at first a dull ache in the left iliac fossa. It got worse, and spread over the lower abdomen radiating down the left leg. A moderately severe diarrhoea developed, with 5 loose stools during the previous 24 hours. Urinary symptoms were absent. On admission the temperature was 38,5°C, the pulse rate 110/min, and the respiratory rate 24/min.

Physical Examination

There was generalised tenderness below the umbilicus, more marked on the left side where an ill-defined mass could be palpated extending down into the pelvis.

Inspection of the perineum revealed an enlarged clitoris, resembling a short penis, 4 - 5 cm in length and 2,5 cm in diameter. It was notched on the ventral surface as for a urethral opening, but the actual urethra opened on the perineum 3 cm lower down. There were two prominent scrotal folds extending downwards to the anus. Anteriorly they were separate as far as the opening of the urethra, fusing posteriorly in a prominent median raphe, thus simulating an empty scrotum. No testicles could be palpated externally.

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Date received: 14 March 1973.

On rectal examination no prostate could be palpated but a diffuse mass, very tender to the touch, was palpable on the left side.

General examination demonstrated the body configuration of a well-developed female with feminine features with well-shaped, round, firm and particularly muscular extremities. There were two well-placed maidenly mammae with adequate nipples. The face was smooth and hairless, and there was a typical female distribution of pubic hair. The rest of the physical examination was negative.

The patient was admitted for observation as a case of acute abdomen. Early the next morning a repeat general and rectal examination showed no change, and he was prepared for laparotomy.

Operation

A laparotomy was performed through a left paramedian incision. On the left side a large tubo-ovarian abscess distorted the anatomy completely. The abscess was opened and the pus evacuated. Diseased tissue was removed as far as possible, and the necessary biopsy specimens taken.

No other acute pathology was present in the renal areas or in any other part of the abdominal cavity.

During the operation an infantile uterus with normal Fallopian tube, and a round, plump right ovary, were noticed. The outline of the uterus could be traced quite easily, but it was impossible to feel whether the cervix was attached to the vaginal vault. Thickened, inflamed tissue made a detailed anatomical dissection and investigation impossible in this area.

Corrugated drains were extruded from both iliac fossae and the abdomen closed in the routine way. Recovery was uneventful.

Laboratory Reports

Culture of pus and sensitivity tests showed the presence of staphylococci sensitive to penicillin, erythromycin and tetracyclines.

Full blood count, blood urea and electrolytes and fasting glucose were all normal.

Exfoliative cytology of buccal mucosa: 12% of nuclei showed 'Barr' chromatin bodies (the nuclei of the cells from a buccal smear contain a dark spot, the Barr body, when 2 X chromosomes are present).[1]

Chromosome analysis proved a normal female karyotype (46XX).

Histological examination indicated that primordial follicles were present in the ovarian tissue submitted.

Twenty-four-hour urine investigation (908 ml in 24 hours) demonstrated oestrogens 0,7 mg/24 hours; and 17-ketosteroids 5,4 mg/24 hours (normal—males 7 - 25 mg,

females 3,5 - 17 mg). Androgens were not estimated.

X-ray examination of chest and pituitary fossa were normal. A sinogram was done with contrast medium through the opening in the perineum, which showed a contrast-filled cavity in the right pelvis, anteriorly. The contrast medium probably lay in the bladder. There were no radiological features to suggest that it lay in the uterine cavity.

Psychosocial Aspects

During his stay in hospital a comprehensive history was obtained on his sexual and social activities from the patient, who was the second of 4 children, with an older sister and 2 younger brothers.

He grew up as a boy, and apparently experienced no difficulty doing so. He had no schooling and spent his youth herding cattle. His relationship with his parents was normal and he got on well with his sister and brothers. He suffered only minor maladies as a child.

He urinates like any normal male and he has never experienced trouble performing the act of micturition. Blood was never noticed in the urine, nor did he ever suffer from discomfort or pain in the lower abdomen. Bowel movements were normal and diarrhoea was never a problem, nor did he ever notice blood in his stools.

He was found to be of a friendly disposition and very even-tempered, but extremely sensitive about his enlarged breasts and he had guarded this secret from the age of puberty. He keeps them covered with a vest or other piece of attire, and avoids bathing in public. He prefers male attire and enjoys farm work, especially building houses, driving and repairing tractors.

He enjoys alcoholic drinks in moderation and his association with his fellow-workers has always been very cordial and friendly. Co-operation with his White employer has always been satisfactory. Socially he is equally at home with either sex, and he felt quite at ease in the overcrowded male ward in which he was nursed.

In 1964 he married and is very fond of his wife, their relationship having been very satisfactory. According to him, their sex life has been without problems, and he enjoys sexual intercourse. His sexual instincts appear to be quite normal. He had confidence in himself and a discussion of the commoner sexual deviations among males did not bring forth any response. One child, a boy now 8 years old, is believed by him to have been born out of this marriage. He would not enjoy being pregnant, nor giving birth to a baby. His relationship with his wife is stable—both physically and emotionally satisfying. He has never had an interest in any other woman.

His wife, an albino woman in her late twenties, was also interviewed. She gave a history of having married this patient 8 years ago. It can be truly said that somewhere there is a lover for everyone, handicap and all, who has the confidence to find her.[2]

He made love to her in the normal way, was very helpful, protective, always trying to please her, but also extremely jealous. She never inquired into his youth and the way he grew up, nor was she interested in his family history. He paid 9 'live' oxen as *lobola* for her, and she feels that for this reason she has to be faithful to him.

Sexual intercourse with the patient gives her no satisfaction. He has poor erections, very poor penetration and no ejaculations. She has never noticed anything suggestive of menstruation on his clothes. He has never undressed in her presence and always keeps a vest on when washing or dressing.

He is very suspicious of other men, but she does not know whether he is aware of the fact that she has a boy friend whom she occasionally meets 'in the bush' and who is the real father of their 8-year-old son. His general behaviour is that of a male. He enjoys farm work and is a good worker, careful about his dress and his behaviour. For recreation he enjoys Zulu dances, drinks moderately and enjoys mixed company. It is only occasionally that he becomes temperamental and emotional.

The albino woman became very emotional during the interview and started crying. Her explanation of this conduct was that she was rather frustrated and depressed.

DISCUSSION

Intersexuality

Intersexuality can be defined as a condition of imperfect sexual differentiation into either male or female. It is a relative term, because no human being is completely male or female. Indeed, the borderline between a normal and abnormal degree of intersex is vague and impossible to define. There is much to be said for the view that femininity is a neuter state and that masculinity is a superimposed characteristic.[3]

The sex of an individual cannot be judged by any one feature, not even the chromatin pattern in the nuclei, the number and type of sex chromosomes and the histological characteristics of the gonad. More important than these, both to the individual and to the community, are the secondary sex characteristics, such as external genitalia, breasts, voice and facial hair. It follows that terms such as male and female pseudohermaphroditism should be abandoned in favour of the non-committal intersexuality'.

The classification of states of intersex is difficult because so many influences, operating before and after birth, are concerned in sex determination and differentiation.

This type of ambiguity is produced in genetic females by the presence of too much masculinising hormone at a critical period of fetal development. This masculinising hormone is produced in the adrenogenital (XX) syndrome by over-active, erroneously-working adrenal glands. In this condition the adrenal cortex makes the wrong hormone—androgen instead of cortisone. The basic cause is a recessive genetic trait. It is the commonest cause of female intersex, and can be controlled by cortisone replacement treatment.

The other type of female intersex is produced by abnormal amounts of androgen from the mother. The mother may have had an androgen-producing tumour while pregnant. The greater likelihood, however, is that she was prescribed progestin to prevent a threatened miscarriage. In a few instances during this treatment the

body utilises the progestin as if it were androgen, so that a female fetus occasionally becomes masculinised.

The metabolism of one hormone into another within the human body is not surprising, since biochemically the sex hormones are all related.

Today the treatment for female intersex—surgical and hormonal—is effective.[4,5] Most important is early diagnosis, ideally at the time of birth. Then the girl can grow up oblivious of ever having had a problem. Her need for special sex education is then correspondingly minimised.

Embryologically the external organs are the last of the sexual morphology to be completed. Nature takes identical initial structures, converting them into either male or female genitalia. The genital tubercle grows out to become a phallus. The skin of the scrotum which also fuses in the midline is the homologue of the labia majora. A median perineal raphe is a male characteristic.

It is relatively simple in such biological engineering for the external genitals to be left unfinished, neither fully masculinised nor feminised and looking remarkably alike.

There is a genital tubercle which could be mistaken for a small penis. This organ has an open gutter underneath instead of a covered urethral tube with an orifice at its root or base, more or less in the female position. The opening may be small and lead directly to the bladder, or it may be quite a large urogenital sinus from which can be traced both the urethral and vaginal passages. The latter may either connect with the cervix of the uterus or end blindly (as in this case). The outer appearance may be ambiguous and thoroughly confusing as to the actual sex. The dilemma in intersex is that of ambiguity of the external appearance.

There arises a rather delicate counselling situation if a female intersex, living as a boy, develops breasts at puberty and begins to menstruate as a result of the oestrogenic activity of hitherto unsuspected ovaries. The best procedure in sex education and counselling is not creating emotional indigestion by saying too much too soon, and also not allowing emotional malnutrition by saying too little to late. Explanations could be based on the child having been born sexually unfinished—an extraordinarily useful term that has spared many patients and their parents the mortification of terms such as 'freak' and 'morphodite'.

In most such cases of adolescent incongruity the patients' feeling and conviction of sexual identity will be in accord with the sex of rearing, the secondary sex characteristics, and especially the appearance of the external genitalia. Masculine trends in outlook and behaviour are often of psychological origin. For some reason or another there is a conscious or subconscious determination to suppress or deny the female sex and this sometimes amounts to overt transvestism and trans-sexuality.

The ovaries, although normally formed, do not function; the uterus remains infantile and fails to menstruate. As the years go by, masculinity becomes so intense that in the days when effective treatment was impossible many sufferers from the adrenogenital syndrome found it easier to live as men.

Providing the diagnosis is made early, postnatal development can be corrected. It requires only a relatively small dose of cortisone or one of its derivatives, determined by clinical trial and controlled by hormone assays in each case, to inhibit the ACTH output and to allow the function of the reticular layer of the adrenal cortex to return to normal.

The abnormality of the vulva can readily be corrected by plastic surgery. Exposure of the vagina can be deferred until puberty to avoid psychological reaction. The large phallus should be removed before the age of 18 months.

Early diagnosis (i.e. excessive amounts of 17-ketosteroids and pregnanetriol in the baby's urine) and treatment are important, otherwise voice changes and premature closure of epiphyses leave permanent stigmata.

Without treatment these girls never menstruate or conceive. With cortisone therapy, which has to be continued throughout life, ovarian and uterine functions become normal.

Vaginograms can be obtained by running radio-opaque fluid through a catheter inserted into the urinary opening. In this particular case great difficulty was encountered in introducing a fine catheter into the urinary orifice. This accounts for the indefinite radiological findings already discussed. Endoscopy might have been helpful. The partly-stenosed urinary canal, making catheterisation so difficult, could be held responsible for the tubo-ovarian abscess which brought the patient to the hospital for treatment. Residual urine in the cavity of the vagina, becoming infected with staphylococci, could have caused an ascending infection through the uterus to the left tubo-ovarian tissues.

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