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CASE REPORTS

Anatomical sex conversion in a 21-year-old — case report and review of literature

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SUMMARY

A case of anatomical sex conversion in a 21-year-old genotypic male Nigerian with ambiguous genitalia but who was reared as a girl is presented. The clinical features observed in this particular case and the result obtained after surgery, adjuvant hormone therapy and supportive psychotherapy are discussed. The relevant literature pertaining to the clinical entity is reviewed.

INTRODUCTION

An individual's social sex (the sex of rearing) and the subsequent legal sex is determined by many factors. These factors are chromosomal, gonadal, hormonal and psychological.

Genetic sex is determined at fertilisation but gonodal sex remains undifferentiated until the 6th week of intrauterine life.[1] Genetic sex usually determines the gonadal sex by permitting differentiation of the indifferent embryonic gonad into either a testis or an ovary.

A normal testis or ovary is a prerequisite for normal sexual, internal and external, differentiation. Thus, the sexual differentiation of the sex ducts — Wolffian and Mullerian — and of the external genitalia is dependent upon the proper endocrine function of the developing gonad.[2]

So, although the facility with which a subject is usually labelled as either male or female is taken for granted, there are a few cases in which this is very difficult. Such cases present with ambiguous phenotypic indices, especially the external genitalia. To these cases, the term intersex has been applied.

Many babies with ambiguous genitalia have had the wrong sex assigned to them and if left uncorrected in early childhood, later present a most difficult problem in management.

Mainly to avoid confusion and psychological disaster, many studies of cases of ambiguous genitalia have recommended that such infants should be thoroughly assessed within the first 12–18 months of life and that a firm decision should be reached on the sex most suitable for the child.[3,4]

Surgical techniques and hormone therapy are then applied to bring the reproductive structures in line with the assigned sex. furthermore, the gonads of these patients usually undergo varying degrees of dysplasia with time. The possibility of development of gonadoblastoma is a major concern — emphasising the need for early surgical assignment of sex in patients with ambiguous genitalia.

Unfortunately, as a result of an absolute lack and an inequitable distribution of specialised manpower, especially in the developing world, older children and adults are sometimes seen in whom the more appropriate sex was not correctly chosen in infancy.

A decision would usually have to be taken in such cases as to whether the wrongly assigned sex should be changed or whether the reproductive structures should be brought in line with the assigned sex by means of surgery and hormone therapy — a kind of 'anatomical sex' change.

Generally, in older children and in adults, the sex of rearing should not be changed, despite a previous illogical judgement, unless in-depth psychological analysis reveals a definite gender identity discordant with the sex of rearing.[5]

CASE REPORT

A 21-year-old supposed female with primary amenorrhoea and ambiguous genitalia was seen, for the first time, by us in 1987. In the previous three years, she had been attending the Gynaecology Clinic where she was being investigated as a case of intersex, having been referred there by a doctor-relative of hers.

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While still being seen by the gynaecologists, she presented to the Accident and Emergency Department with acute abdomen and was subsequently referred to us as a case of strangulated left inguinal hernia.

The patient gave a one-day history of sudden onset severe left inguinal pain associated with a swelling of the left 'labial' region. 'Female circumcision' had been carried out during childhood and she had had appendicectomy at the age of 15 at a General Hospital. She was told at the General Hospital, then, that she would require further surgery to correct hepambiguous genitalia.

She was the second of eight children in the family. There was no family history of ambiguous genitalia. Her immediate younger female sibling, 15, had already achieved menarche. She has had suicidal tendency since puberty when she found she could not menstruate like her peers.

On examination, she was a skinny, 162cm tall subject. She had broad and squared shoulders, facial hair, a receding frontal and temporal hair line and a deep male voice. She weighed 24kg. the breasts were under-developed but she had 'female pattern' pubic hair. She had a peniform 'clitoris' which was ensheated in infantile scrotolabial folds. A tender firm mass about 3cm x 5cm was felt in the left groice but there was no mass on the right. The introitus to the 'vagina' admitted only the tip of one finger.

A poorly defined urethral meatus was located in the posterior aspect of the introitus and the 'vagina', shallow, ended blindly. Bimanual examination revealed no uterus but a prostate was demonstrable on rectal examination. Her cardiovascular status was normal and she had o skeletal abnormalities.

She had a haemoglobin of 10g/dl. Serum electrolytes and urea were normal. A previous buccal smear examination revealed that less than 2 pc of the cells contain Barr bodies. A full chromosome analysis was not done. Ultrasonographic examination showed absent ovotestes and no uterus. Intravenous urogram was normal. There was no facility for hormonal assay at the time of presentation.

Emergency exploration of the left groin mass revealed a gangrenous testis which had undergone torsion, associated with a small indirect inguinal hernia. Orchidectomy and herniorrhaphy were performed. Histopathology confirmed the gangrenous mass to be testicular.

Figure I: Patient's profile before anatomic sex conversion. Note the striking male features.

Figure II: Eighteen months after anatomic sex conversion.

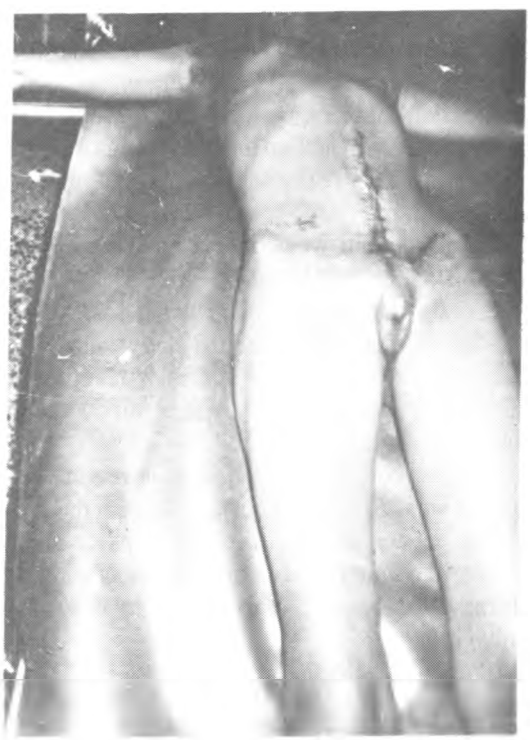

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Figure III: Examination under anaesthesia at 18 months after surgery. Note the breast development.

Since the patient expressed her wish to remain a 'female', she was readmitted six weeks later for the purpose of complete orchidectomy and vaginal contraction. Exploration for the right groin revealed a testis arrested in the line of descent. This was removed. The right inguinal canal was then obliterated.

A neovaginum was constructed with a pedicled sigmoikd colon transplant.

Post-operatively and commencing on the 8th day, the neovaginum was dilatated daily with Hegar's dilators. From then on, she also received oral stilbesterol 5gm thrice daily, which was later stepped up to 10mg t.d.s.

The post-operative period was uneventful and the patient was discharged home on the 16th day.

The patient also benefited from psychotherapy. Twenty days after the commencement of stilbesterol therapy, the breasts were noticed to have started enlarging.

Two-and-a-half years after surgery, the vagina van admit two fingers, the breasts are still enlarging, the temporal hair recession has been halted and the prostate is regressing in size.

In addition, the patient appears to be adjusting well, psychologically, bearing in mind that menstruation and fertility are not possible. She, however, enjoys an active sexual life. With the absence of testis and astrogen therapy, the prostate gland has involuted completely.

DISCUSSION

The aetiology of ambiguous genitalia include true hermaphroditism, male pseudohermaphroditism, female pseudohermaphroditism, maternal ingestion of hormones, mised gonadal dysgenesis and the Stein-Leventhal syndrome.[5,6]

In true hermaphroditism, the individual has a mosaic of 46,XY and 46,XX chromosomes in about equal proportion, and possess both ovarian and testicular tissue. Female pseudohermaphroditism (46,XX) includes congenital adrenal hyperplasia in which there is usually an enzyme deficiency e.g. of 21 hydroxylase or 11-beta hydroxylase.

Male pseudohermaphroditism (46,XY) includes Delection syndromes with Y cell lines (45,X46,XY), agonadia, various enzyme deficiencies (e.g. 17-Ketoreductase, 5-alpha reductase, 17 alpha-hydroxylase), testicular feminisation (androgen insensitivity) syndrome and the non-endocrine/non-chromosomal defects.

Although our patient's hormonal and enzymatic profile was undetermined, her clinical features were similar to those of male pseudohermaphroditism due to steroid 5 alpha Reductase deficiency as described by Imperato-McGinky *et al.*[7] Inheritance of the enzyme deficiency is autosomal recessive; which means there is a 25 pc risk for recurrence in subsequent pregnancies. This enzyme mediates the conversion of testosterone into its active form — dihydrotestosterone.

Clinical studies have suggested that the differentiation of the external genitalia in the male foetus is effected through the conversion of testoasterone to dihydrotestosterone. Without this conversion, external genitalia naturally develop along female lines.[6]

In male pseudohermaphroditism due to steroid 5 alpha Reductase deficiency,the child has a female phenotypic appearance, but the post-pubertal phenotypic appearance — virilisation, deepening of the voice, an increase in muscle mass — is typically male. This may have been an added deciding factor with regard to the wrong assigned sex of our patient.

The management of this patient involved a combination of surgery, hormonal therapy and psychotherapy. The line of management was determined by the gender role and identity she had assumed. The chromatin study clearly indicated her genetic male status. She had positive chromatin material in less than 2 pc of her cells, unlike the 31 pc obtained in normal females.[8] This was further confirmed by histological examination of the gonads.

That our patient had assumed a female gender role is not surprising as gender role (i.e. the individual's conviction of being male or female) is more in accord with the sex of rearing than with either the kind of gonad or the predominant appearance of the external genitalia as has been concluded from a number of clinical studies.[3]

In constructing a vagina for this patient, we chose a sigmoid transplant because, although it involves more major surgery, it lacks the tendency of the vagina constructed by the purely perineal procedures that may undergo subsequent contraction and the results are very satisfactory.[9]

In addition, the mucus produced by the neovaginum provides good lubrication to facilitate intercourse. It is significant that two years after surgery, our patient has retained a commodius vagina and has had a reasonable sexual life.

In the absence of varies in this patient, we placed her on stilbesterol to effect a female habitus. fortunately for her, she is not a candidate for endometrial carcinoma which has been reported after stilbesterol therapy in gonadal dysgenesis.[10]

On the other hand, she is also excluded from menstruation. A complication of prolonged estrogen therapy, among several, is deep vein thrombosis. Our patient has developed calf pains. she has therefore been placed on dipyridamole and aspirin.

As has been pointed out, cases of ambiguous genitalia that undergo corrective surgery at an early age are psychologically better adjusted than the late cases. This patient presented late, but fortunately, she elected to retain her assumed gender and with the aid of psychotherapy her psychological status has remained satisfactory. She no longer has suicidal tendency.

Early surgery would also help to avoid the development of torsion of the testes as occurred in this patient and, therefore, enable those patients whose parents would rear as boys to retain viable gonads.

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