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ORIGINAL CONTRIBUTIONS

Management of Intersexual Disorders in Infancy and Childhood*

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Aimakhu, Vincent E. (1974). *Nigerian Journal of Paediatrics*, 1 (2), 51. **Management of Intersexual Disorders in Infancy and Childhood.** In a two-year period, 22 cases of intersexual disorders in infancy and childhood were encountered. A majority of the cases were female intersexes. Most of the cases were seen after the age of one month, and some of the reasons for this delay in seeking medical attention include parental feeling of shame, or the desire to have a child with a particular sex, and casual examination of the baby at birth by midwives or doctors.

Surgery which is the principal form of management of these intersexual disorders has been undertaken in 14 of the 22 patients. The anatomical anomalies of the external genitalia were successfully corrected and the immediate results have been encouraging. Parents have also accepted the results with satisfaction.

INTERSEXUALITY (synonym, Ambiguous external genitalia, AEG) may be defined as a condition of imperfect sexual differentiation into either male or female sex. The condition commonly manifests, and, is usually first noted in the external genitalia.

No satisfactory clinical classification of intersexual disorders has yet been devised. Attempts to classify these disorders on the basis of anatomical defects, histology, or sex hormone production have lead to confusion since there is a great deal of overlapping between the various clinical types. However, on the basis of the appearance of the external genitalia, cytogenetic studies, and the type of gonads, we have adopted the following clinical classification (Table I) which is simple and of practicable value.

TABLE I

Clinical Types of Intersexual Disorders

Type	*Sex*		*Gonads*
	Chromatin	Chromosome	
Female Intersex (masculinized female)	Positive	XX	Ovaries
Male Intersex (undermasculinized male)	Negative	XY	Testis
Hermaphroditism	Positive/ Negative	XX/ XY mosaics.	Ovotestis

The child with uncertain sex is a source of grave anxiety and embarrassment to the parents as well as to the child when it grows up (Gordon and Dewhurst, 1962; Dewhurst and Gordon,

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1963). In order therefore to minimize the anxiety in the parents and also to prevent permanent psychological trauma to the child when it grows up, early detection and management of the condition is desirable, since change of the assigned sex at puberty, even though occasionally possible, (Berg, Nixon and MacMalion, 1963) is usually difficult, (Money, Hampson, and Hampson, 1956).

This communication reports our early experience in the management of cases of intersex in infancy and childhood at the University College Hospital (UCH), Ibadan.

Patients and Investigations

Between January 1971 and December 1972, 30 patients with AEG were referred to the Human Cytogenetic Unit at the UCH, Ibadan. Four of these patients were indeed cases of hypospadias, and another 4 were adults, aged between 18 and 35 years, with intersex. These eight cases have been excluded from this series.

There were therefore 22 children and they form the basis of this report. The age when they first sought medical attention is presented in Table II. It will be observed that 14 of the 22 patients (63.6 per cent) sought medical attention during the first year of life. The clinical types of the disorder in the 22 children are summarized in Table III. It will be seen that female intersex was the commonest type.

TABLE II

Age when 22 Children with AEG First Presented

Age	*No. of Cases*
Month	
1	2
1–3	6
4–6	3
7–9	1
10–11	2
Year	
1–2	4
3–4	3
5–6	1
Total	22

TABLE III

Clinical Types of 22 Cases of Intersex

Type	*No of Cases*
Female Intersex	11
Male Intersex	7
Hermaphrodite	4
Total	22

A full clinical history of each patient including the family history was obtained. Enquiry into drug administration to the mother during pregnancy was also made. A complete physical examination of the child and the mother was then carried out, paying particular attention to the presence or absence of any ovarian tumour in the latter.

Investigations carried out on each patient included all or some of the following:

(*a*) Cytogenetic investigation (buccal smear staining, flourescent microscopy and chromosome karyotype).

(*b*) Endocrinological investigation (urinary 17-ketosteroids, urinary pregnanetriol and plasma cortisol and or pregnanetriol)

(*c*) Contrast radiography (vaginogram, pelvic pneumogram and intravenous pyelogram).

(*d*) Genetic studies, (glucose-6 phosphate dehydrogenase (G-6-P-D) activity, rhesus factor and haemoglobin genotype).

(*e*) Serum electrolytes and urea.

(*f*) Laparoscopy and exploratory laparotomy.

Management

Although the method of management varied from case to case, the principles underlying the decision taken on each case were based on the following concepts:

1. Cases of female intersex should always be managed as females regardless of the condition of the external genitalia.
2. All other cases of intersex should be managed according to the sex which offers them the best chance of normal sexual relations later regardless of the nature of the gonads.
3. Everything possible must be done during the first few years of life to *emphasize the decision taken.* This involves appropriate endocrine or surgical treatment to ensure that the existing manifestations of the unwanted sex and the appearance of others, at some later time, are prevented, and, deliberate indoctrination of the parents that their child is a boy or a girl.

Fourteen of the children have been operated on so far. Three cases each representing the three common types of intersex are presented to illustrate the above principles of management.

Illustrative Cases

CASE I (Female Intersex): J. O., (UCH, No. 265792) aged one year was referred from the paediatric department with the main complaint of ambiguous external genitalia. The mother gave the history that even though she thought that the child was a male, 'he' was not passing urine though a 'penile' orifice. Rather the orifice through which the baby passed urine was below the 'penis'.

The patient was the last of four children, the other three, two girls and one boy were perfectly normal. The only drugs taken by the mother during the antenatal period were haematinics and antimalarials.

On physical examination, the only abnormalities were in the external genitalia. There was an enlarged phallus (Fig. 1a), the glans of which was not covered by a prepuce. The phallus was not transversed by a urethra; instead, there was a urethral meatus just at the root of the phallus and this meatus was partly covered by a high perineum. No vaginal opening could be visualized. The labial folds did not contain any gonads,

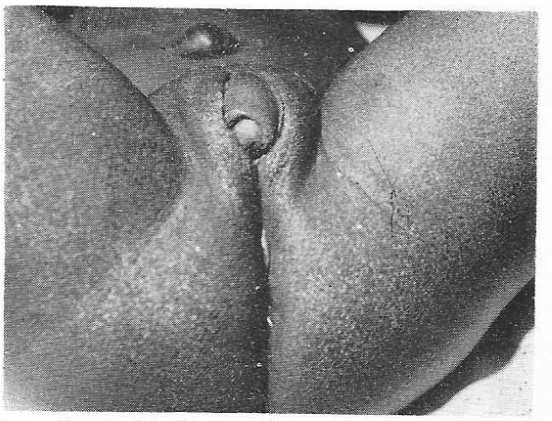
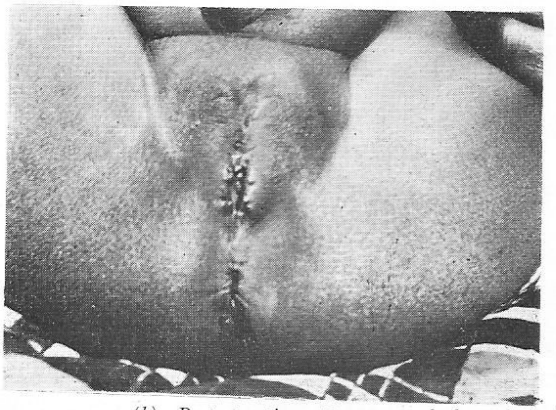


Fig. 1. Case I (a) Pre-operative appearance of the external genitalia of a case of female intersex. Note the enlarged clitoris and high perineum.



(b) Post-operative appearance of the vulva of same patient.

and there were no inguinal hernias. A clinical diagnosis of a female intersex was made.

Investigations revealed 25 per cent chromatin positive cells in the buccal smear. There was a normal sized single Barr body in each cell. There were 46 chromosomes with XX karyotype in all the cells examined from peripheral blood cultures. There was no evidence of mosaicism or X-chromosome abnormality. The plasma cortisol levels were 49 micro-gram (10.00 a.m.) and 35 micro-gram (12 midnight) per 100 ml. These values are within the normal range for a normal female infant (Adadevoh, B. K.—personal communication). The 17-keto-steroids level in a 24-hour specimen of urine was 0.6 mg (normal

value for children over one week old is 1mg./24 hours—Dewhurst and Gordon, 1969). The serum electrolytes and blood count revealed normal values. G-6-P-D was non-deficient. An intravenous pyelogram revealed a normal urinary tract.

Under general anaesthesia, the enlarged phallus was amputated at its base and the mons pubis reconstituted. When the perineum was

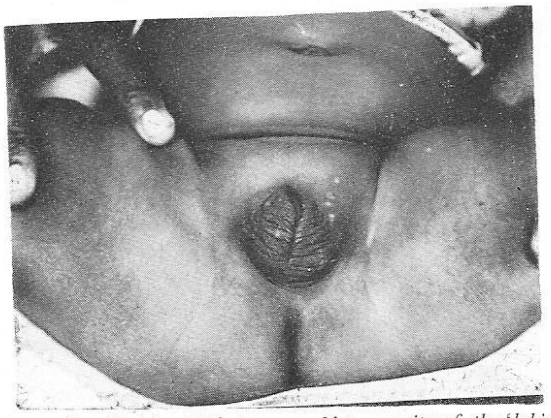


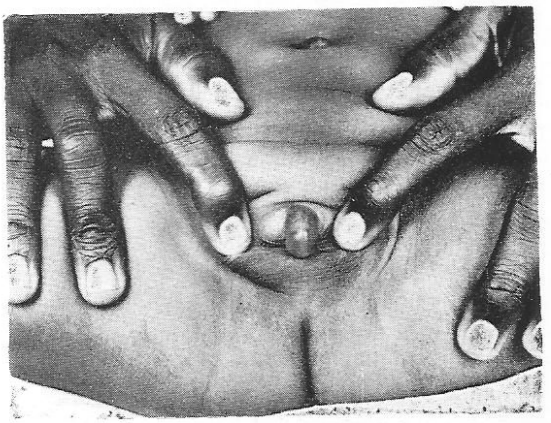
Fig. 2. Case II (a) Male intersex. Note rugosity of the 'labia' looking like scrotal sac.

divided in the mid-line, a vaginal opening was revealed. A vulvovaginoplasty was then performed. A small polythene tube was inserted into the bladder through the urethra for continuous bladder drainage.

Post-operatively, the patient received a five-day course of penicillin and streptomycin. The polythene tube was removed on the fifth day. The post-operative period was uneventful. The histology of the amputated phallus was reported to be consistent with a clitoris. Fig. 1(b) shows the post-operative appearance of the external genitalia. The patient is being reared as a female, and her mother is very satisfied with the result.

CASE II (Male Intersex) W. A., (UCH, No. 300172) was first seen at the age of five months. The parents, who were obviously very anxious about the child's sex, complained that, because of the baby's abnormal external genitalia (Fig. 2a), they could not decide on its sex.

On physical examination, the external genitalia consisted of what looked like vulval folds, with marked rugosity very suggestive of a bifid scrotal sac. The 'folds' contained very small gonads, and on separation of the 'folds' a small phallus was revealed. This phallus was not transversed by the urethra; rather, a urethral meatus was situated below it. The glans penis was not covered by a prepuce (Fig. 2b). A clinical diagnosis of a male intersex was made.



(b) Separation of the 'labial' folds exposes a small phallus with no prepuce covering the glans.

All cells in the buccal smear were chromatin negative; there were also 46 chromosomes with an XY karyotype in all the cells in the peripheral blood cultures. Serum electrolytes, urea, haemogram, and intravenous pyelogram were all normal; G-6-P-D was non-deficient.

It was explained to the parents that the child was not completely differentiated into a male and that it would be best to rear the child as a female. The parents were further informed that the construction of the vagina surgically and stimulation of breast development with drugs would be carried out when the child was older. The phallus was amputated under general anaesthesia, and bilateral gonadectomy performed. The ears were pierced. The gonads on histology revealed immature testes. The child is being followed up at yearly intervals. The

parents are satisfied with the decision regarding the sex and are very pleased with the results of the operation.

CASE III (*Hermaphrodite*) A. O., (UCH, No. 282603) was initially referred to the consultant urologist at the age of 18 months as a case of an undescended left testis because the right gonad was palpable in the scrotum (Fig. 3). At operation to correct the defect, the urologist found a uterus, fallopian tubes and an ovary on the left side of the pelvic cavity. It was at this stage that gynaecological opinion was sought.

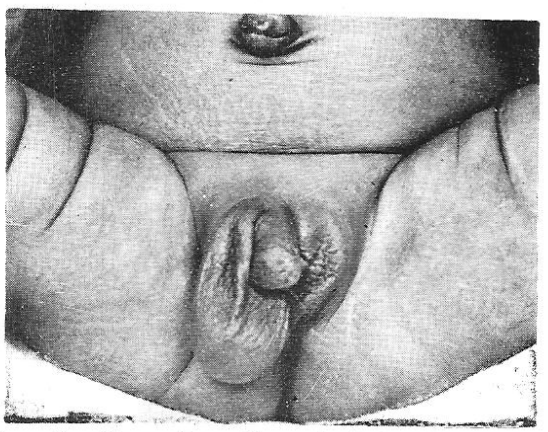


Fig. 3. Case III: A case of hermaphrodite initially referred as a patient with undescended left testis. Note the well formed phallus and right scrotal sac containing a gonad.

Investigation showed six per cent positive chromatin cells in the buccal smears, which was suggestive of mosaicism. But all the cells analysed in peripheral leucocyte cultures has 46 chromosomes with XX karyotype. Since the phallus transversed by a urethra was of a reasonable size, it was decided to rear the child as a boy. Consequently, total abdominal hysterectomy with removal of both the gonads and fallopian tubes was performed. Pellets were made from stent mould and left inside both scrotal sacs to simulate testes. The gonad in the right scrotal sac was indeed an ovo-testis.

The parents, who already had four daughters and wanted a male child were pleased. It was, however, explained to them that the child would be sterile because of the developmental anomaly. In later years the child will be given testosterone to establish and maintain male characteristics.

Results of Management

Our immediate results have been encouraging. First, there has been no surgical mortality. Occasionally, however, vulval haematomata occurred. Secondly, most parents have accepted the explanation given for the decision taken on the sexing of the children.

It is too early at this stage of our experience to express an opinion on the long term results in respect of the male intersexes that have been converted into females. With regard to the female intersexes, the long term results are expected to be good, since fertility is usually not impaired.

Discussion

Intersexual disorders are not uncommon in Nigeria. In technically developed countries, where most babies are delivered under the supervision of trained personnel, most cases of intersexual disorders are diagnosed at birth or soon after. Management of these disorders in the newborn period minimizes the period of parental anxiety, and also the problems of the older patients with uncertain sex.

From the present study it can be seen that in most cases of intersex, parents seek medical attention rather late. The reasons for this may include: delivery of babies by non-medical/nursing personnel; casual examination by midwives or doctors of the external genitalia at birth; parental feeling of shame or the desire to have a child with a particular sex. In the present series, two cases of parental desire for a particular sex was the cause of delay in seeking advice. Indeed, the parents of one of these two children have rejected the diagnosis of female intersex.

Although most of the cases were seen after the age of one month, most parents had fortunately not decided on the sex of rearing of their children. This delay on the part of the parents in deciding

on the sex of rearing of their children, made it possible for them to be managed as if they were newborns.

Dewhurst and Gordon (1969) have reported that female intersex is more common than the other types. In the present study there were more cases of female intersex (50 per cent) than the other types. In contrast to the experience of other workers no case of adrenogenital syndrome was encountered among the cases of female intersex. It is possible that some cases of adrenogenital syndrome (particularly the salt-loosing type) in the environment die early due to the delay in seeking medical treatment. It is also possible that the masculinization of the external genitalia in some cases may be due to progestational substances in traditional herbs taken by mothers during pregnancy.

The management of the newborn infant with intersex is basically a decision on sex rearing. This decision is usually the joint responsibility of the child's parents and the gynaecologist. Fortunately in the present series no psycotherapy on the older children was necessary, probably because the sex of rearing had not yet been decided upon by parents before medical consultation.

Surgery is the mainstay of definitive management. With regard to female intersex, all gynaecologists believe in amputation of the phallus and performing a vulvovaginoplasty. There is unanimity on early amputation of the phallus, but some people would defer the vulvo-vaginoplasty until the child is older. It is our practice to amputate the phallus and perform vulvovaginoplasty at the same time since one cannot rely in this country on the parents to bring back their children, once the phallus (the main source of embarrassment) has been amputated.

As for the male intersexes converted into females, bilateral orchidectomy is the initial procedure. Vaginoplasty in these cases is, however, deferred until the child is much older for two reasons: First, these patients will almost certainly come back later because of primary amenorrhoea. Secondly, the procedure of vaginoplasty in these patients is not only different from that in the female intersexes, but it is also easier to perform when the child is older and the tissues are more developed. In addition to surgery, oestrogen therapy in these patients should be started around the age of 12 years, in order to initiate and sustain breast development. It is our practice during orchidectomy to pierce the ears of these children (if this had not already been done) as a deliberate attempt to impress on the parents that these children are girls.

The immediate results have been good. The anatomical anomalies of the external genitalia have been successfully corrected and the parents have been pleased. The long term results should be good especially in the female intersexes, whose fertility is not expected to be affected. Finally and probably most important, by the early diagnosis and management of these cases, the psychological problems of the older patient with uncertain sex is prevented.

Acknowledgements

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