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SURGICAL MANAGEMENT OF NEONATES AND CHILDREN WITH AMBIGUOUS GENITALIA

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Gender assignment is a neonatal surgical emergency. Early evaluation and treatment of intersexuality must be done as a team approach with a genetist, a paediatric endocrinologist and a paediatric surgeon/urologist taking part in it. Change of gender after 2 years of confusion of gender identity often results in social disaster.

Over a period of 20 years the authors managed a total of 27 patients with ambiguous genitalia. They favour early reconstruction of most anomalies at about 3 years of age, which seems to be optimal in terms of surgical feasability and avoidance of the obvious emotional problems caused by delayed intervention. A detailed description of policy and techniques is given for surgical intervention in the patient to be raised as a female or as a male. They changed the gender in three patients, twice in neonates and once before two years of age.

INTRODUCTION

With a few exceptions such as the salt loosing form of congenital adrenal hyperplasia the condition known as ambiguous genitalia does not constitute a threat to life but its incorrect evaluation and management may lead to unhappiness for both parents and child. A delay in making a gender assignment of the wrong gender requiring us change is often fraught with emotional trauma and social disaster /3, 5, 8/.

Paediatric surgeons are often involved not only in reconstructive surgery but also in making a diagnosis by clarifying the anatomy in a particular case. Their

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TABLE I

Most important clinical data of the 27 patients

No. of patients	Age at first hosp.	Diagnosis	Chromosomal find. Associated anom.	Surgical diagnostic and therapeutic interventions	Range of ages at intervention (yrs)
1 - 6	neonates	cong. adr. hyperplasia /CAH/	46XX salt losing /1x/	one-stage feminizing genitoplasty /5x/ clitorectomy+flap vaginoplasty /1x/	2 6/12-8 yrs
7	3 mos	clitoral enlargement	46XX	clitoral recession + relocation	3 yrs
8	13 yrs	compl.test. feminization	46XY	laparotomy + gonad biopsy gonadectomy + clitorectomy	13 yrs 13 3/12 yrs /female to male/
9 - 11	neonates	incomplete test. fem.	46XY /2x/ associated anomalies + Meckel Gruber sy 1x	gonad biopsy /3x/, laparotomy /3x/, removal of dysgenetic gonads /2x/, clitoral recession + relocation /2x/, clitorectomy /1x/, flap vaginoplasty /1x/	10 days - 7 yrs
12 - 17	neonates - 7 yrs	mixed gonadal dysgenesis	46XY /3x/ 45X/46XY /1x/ 46XY/45XO /2x/	laparotomy /3x/, chordectomy /3x/, removal of uterus and salpinx /2x/, gonad biopsy /6x/, orchydopexy /4x/, hypospadiasis repair /2x/, change of gender /1x/, clitoral reduction plasty /1x/,etc.	1 mo - 12 yrs
18 - 19	neonate - 2 yrs	true hermaphroditism	46XY 46XX/46XY chimerism	laparotomy /2x/, gonad biopsy /2x/ flap vaginoplasty /1x/, change of gender /male to female/ /1x/	2/52 - 13 yrs
20	2 yrs	Brook pseudohermaphr. masculin	46XY	laparotomy, gonad biopsy	2 yrs

21	neonate	fetal hydatoin syndrome	46XY multiple assoc. anomalies	gonad biopsy, laparotomy, chordectomy, hypospadias repair	5/52 - 3 yrs
22 - 23	neonates	pseudo-vaginal perineoscrot. hypospadiasis + kryptorchism.	46XY	gonad biopsy /2x/, orchidopexy /2x/, hypospadiasis repair /2x/ one of them not yet finished	3/52 - 4 yrs
24 -26	neonates	hyperplasia of labia minora	46XX	removal of redundant labia minora	6 - 13 yrs
27	neonate	penile agenesis	46XY	perineal urethroplasty, gonadectomy, creation of labia minora, change of gender /male to female/	1/52 - 8 mos

participation in the X-ray studies, endoscopy or taking tissue samples will considerably contribute to early and proper diagnosis. After gender assignment the paediatric surgeon/urologist will perform the necessry feminizing or masculinizing interventions which often consist of a sequence of surgical procedures.

This report presents our experience with the surgical diagnostic methods and reconstructive techniques used in the evaluation and treatment of patients with ambiguous genitalia in our department.

MATERIAL AND METHODS

Over a period of 20 years (from 1st January 1968 to 31 Deceber 1987) 27 patients, mainly neonates with amiguous genitalia reguiring either diagnostic or/and reconstructive surgical procedures, were evaluated and treated in our department.

Table I shows the most important clinical data of the 27 patients, 18 of whom were seen initially as neonates at our department and 9 were referred to us for first or further evaluation and/or treatment.

The paediatric surgeon's role in the evaluation of patients with amiguous genitalia

Paediatric surgeons are very frequently actively involved not only in gonad biopsy and laparotomy but also in X-ray studies (genitography and endoscopy).

X-ray studies (genitography)

Conventional cysto-urethrography may be misleading in determining the presence or absence of a vagina within a single perineal opening. Accordingly a "flush genitography" abvocated by Hendren et al. can be performed. This is done by placing a blunt-tip syringe filled with dye in the single perineal opening /6/. We prefer the use of a catheter introduced into the bladder and urrogenital sinus and fixed to the perineum witrh adhesive tape so as to prevent leakage of the dye injected into the sinus (Fig. 1).

Endoscopy

The use of miniature optic endoscopes (Ch 8-10) has been a major breakthrough in the diagnosis and treatment of genitourinary conditions at any age and has proved very helpful in our experience.

We use a cystoscope to determine whether the vagina enters the urethra or urogenital sinus. If the vagina enters distal to the external sphincter a later detailed relatively simple surgical procedure, flap-vaginoplasty may be performed. If the vagina enters the urethra proximal to the external sphincter, then a more complicated pull-through vaginoplasty must be done.

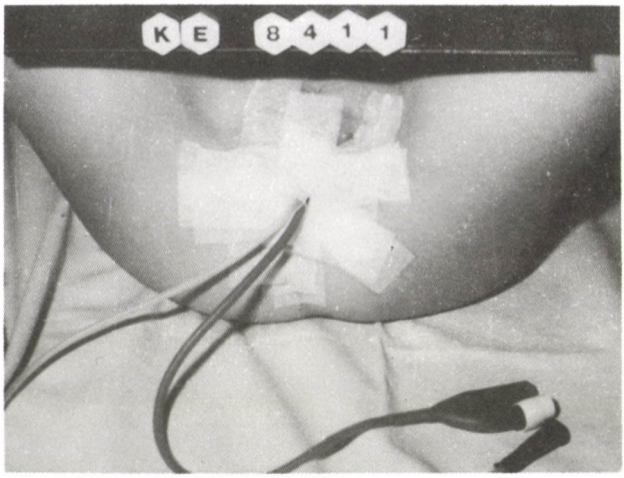

Fig. 1. Genitography: catheters introduced into the bladder and urogenital sinus are fixed with adhesive tape.

Gonad biopsy and/or laparotomy

In a few instances the above-mentioned diagnostic procedures together with a careful physical examination, chromosomal analysis, biochemical testing will not clarify the pathology and will not provide sufficient data for gender assignment and subsequent corrective surgery. In these cases labioscrotal gonad biopsy and laparotomy might provide further important data (Table I). During laparotomy the presence or absence of internal genitalia should be established (uterus, salpinx, ovaries, testes - streak - spermatic cord) and tissue samples taken. In order to detect ovotestes, longitudinal tissue samples should be taken from the middle parts of the ovaries or testes. Exstirpative surgery must not be carried out before knowing the result of gonad biopsy.

Surgery if a child is to be raised as a female

A feminizing genitoplasty should be directed to three goals:

1. Removing the corpora and preserving the glans with its innervation to create a clitoris with normal sensation
2. Creating a normal-appearing introitus by fashioning labia minora from phallic skin and foreskin
3. Vaginoplasty to provide an adequate opening for the vagina into the perineum.

Snyder and associates /11/ have developed an operation for congenital adrenal hyperplasia which incorporates all above-mentioned goals. Since its publication, we have performed this feminizing genitoplasty in 5 patients with congenital adrenal hyperplasia (Types II and III according to Prader - /10/), and this relatively simple procedure gave very good cosmetic results. Considering that we reported about our favourable results with this feminizing genitoplasty in detail elseewhere /9/. We are now supplying only the photos of the

most important steps of this procedure (Fig. 2 a-f).

If female gender was assigned to a patient with mixed gonadal dysgenesis or true hermaphroditism at birth (or later), a feminizing type of genitoplasty consisting of clitoroplasty, and the creation of labia minora should be performed. At the same time, the dysgenetic gonads (streak and testes) are removed because of an increased incidence of malignancy and in order to prevent further virilization at the time of puberty /2/. In patients with true hermaphroditism the gonads may be microdissected and the gonadal tissue discordant with the sex of rearing removed /3/.

We had 6 patients with mixed gonadal dysgenesis; at birth 2 of them were assigned female gender, which was changed in one of them female of male (Table I).

One of two patients with true hermaphroditism was assigned female gender and a feminizing genitoplasty similar to that used vor congenital adrenal hyperplasia wos performed. In the other who was assigned male gender the predominant female anatomy necessitated a change of gender at the age of 20 months /Table I/

Children with complete (familial) testicular feminization syndrome appear as normal phenotypic females with practically no signs of virilization. During adolescence, these children frequently seek attention because of amenorrhea. When discovered, clitoroplasty, labioscrotal reduction and gonadectomy are advised and they are continued in their female role with appropriate hormonal support from puberty on.

Vaginal reconstruction should be delayed until late will provide a vagina of sufficient length or at least an introitus which is less fragile ut oestrogen-responsive /3/ and to which a skin or clon graft can be anastomosed proximally. The patient with complete testicular feminization is now over 20 years and there is no need for vaginal replacement. We had 3 patients with incomplete testicular feminization (dysgenetic male pseudohermaphroditism). They were born with an enlarged phallus and a rudimentary vagina behind a common single perineal opening. Reconstruction was again towards the female gender in all three patients and the dysgenetic gonads, ovotesten or testes were removed /Table I/.

Surgery if a child is to be reised as male

In patients with male pseudohermaphroditism who are supported in the male gender the surgical procedures can be divided into two main groups: exstirpative interventions and creation of an external male appearance.

The first group of procedures includes removal of the gonad tissues and the internal genitalia (uterus, salpinx) discordant with the sex of rearing (Fig. 3 a-b).

Orchidopexia hypospadiasis repair are the two most frequent interventions in patients raised as males /1/.

Orchidopexy can be very difficult since the intraabdominal testes may not only have short spermatic vessels but also short vasa, sometimes making the bringing down of the testes into the scrotum impossible. With transposition of the cord structures

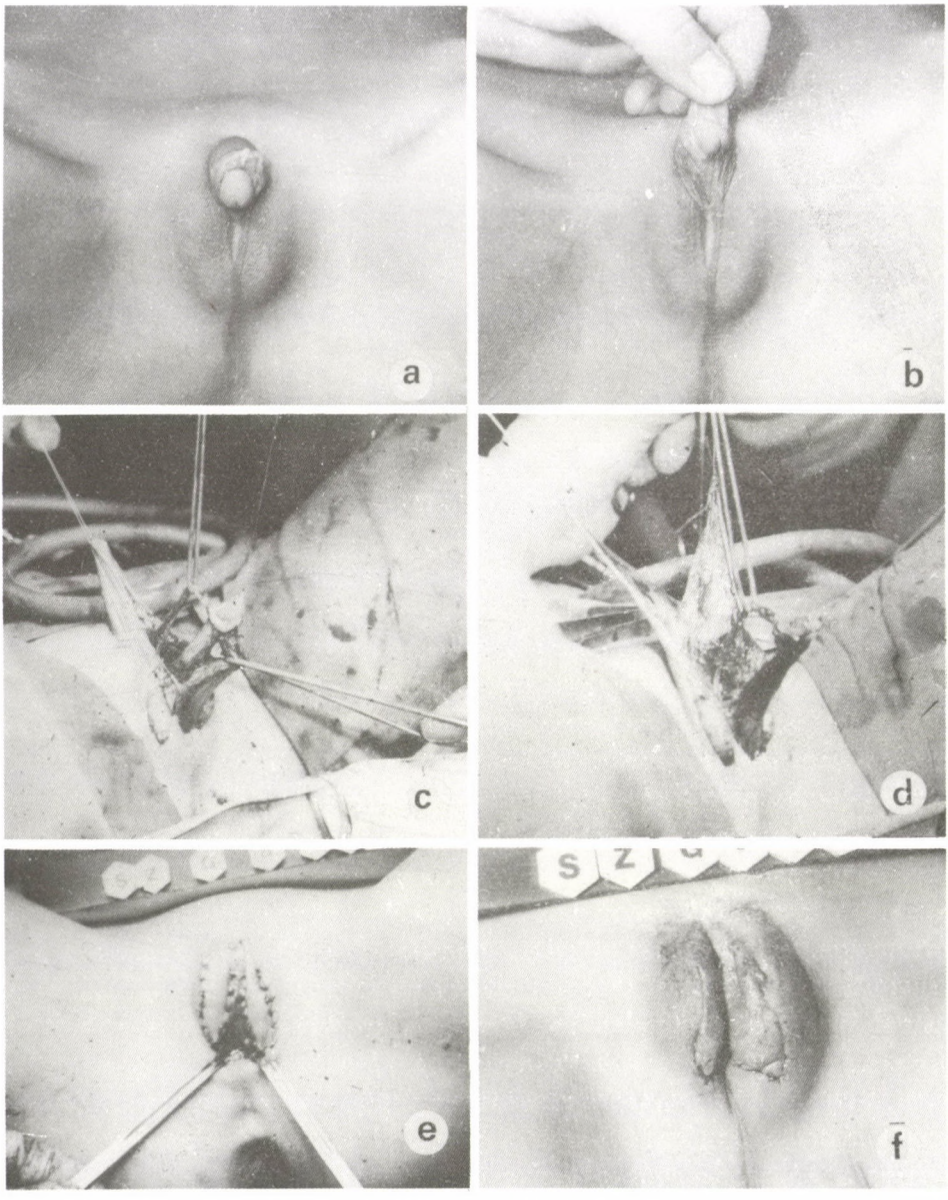

Fig. 2. a-b Enlarged clitoris, opening at its base, fusion of the labia minora.
c Preservation of dorsal innervation and ventral blood supply of the glans.
d After resecting the crura, phallic skin is split in the midline.
e-f Creation of labia minora using the phallic anoforeskin.

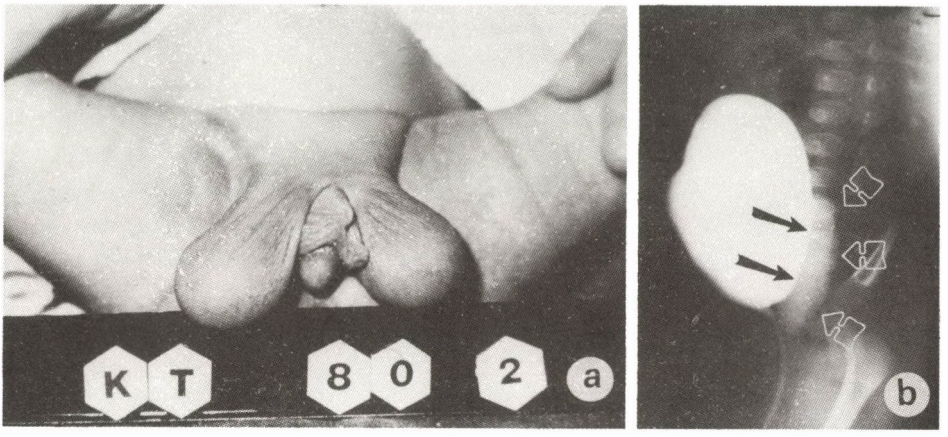

Fig. 3. Neonate with mixed gonadal dysgenesis.
a External appearance
b Genitography: hypoplastic uterus behind the bladder.

medially the course of the spermatic artery may be lengthened effectively and the testes may reach the scrotum. If this is not possible the gonad is left in higher position and six months lates during a second surgery the testes can be mobilized and placed into the scrotum.

Difficult hypospadiasis repair /4/ is frequently required in patients with male pseudohermaphroditism who are to be supported in the male gender. The perineal opening is often at the penoscrotal junction. This together with micropenis and chorda makes hypospadiasis repair one of the most difficult surgical prrocedures. An extended chordectomy will lengthen the penis. The neourethra can be tubularized from the skin of a bifid scrotum, which is joined to a skin graft, either free or vascularized from the dorsal hooded foreskin. If necessary, a neourethra can be constructed from a mucosal graft taken from the bladder, as we did once with good result. The neourethra should then be brought out at the tip of the glans and the glans recreated ventrally to give a normal appearance. In 4 patients (3 with mixed gonadal dysgenesis and 1 with male pseudohermaphroditism) we carried out altogether 4 orchidopexies, 4 chordectomies and 2 hypospadiasis repaire. Two children are still awaiting further reconstruction.

Males with perineal hypospadiasis

Perineal hypospadiasis in the male, which can be regarded as a failure of full virilization, may also manifest itself with apparent ambiguity, especially with bilateral cryptorchism /2/. Two of our patients belonged to this group of children. Gonad

biopsy proved the male gender, orchidopexy and hypospadiasis reepair improved the male appearance.

Hyperplasia of labia minora

In 3 females considerable hyperplasia of the labia minora was responsible for the ambiguous appearance of the genitalia. Removal of the excess of tissue gave a normal appearance to the external female genitalia /Table I/.

Penile aganesis

Aganesis of the penis is an extremely rare anomaly, occurring once in 30 million births. This complex malformation requires urgent assessment at birth for several reasons: female sex assignment is required early. Gonadectomy should be performed in the first few days or months to prevent male gender from developing in response to the testosterone surge occurring between the 10th and 60th days of life. Early gonadectomy and genital reconstruction helps the family to accept the child's altered gender and to reduce phsychological problems /7/. We treated one patient with this rare type of intersexuality (Fig. 4. a-b). At the age of 3 weeks the urethral meatus opening into the lower part of the rectum was transposed to the perineum. At the age of 8 months the patient underwent bilateral orchidoctomy with the scrotum preserved for future viganoplasty.

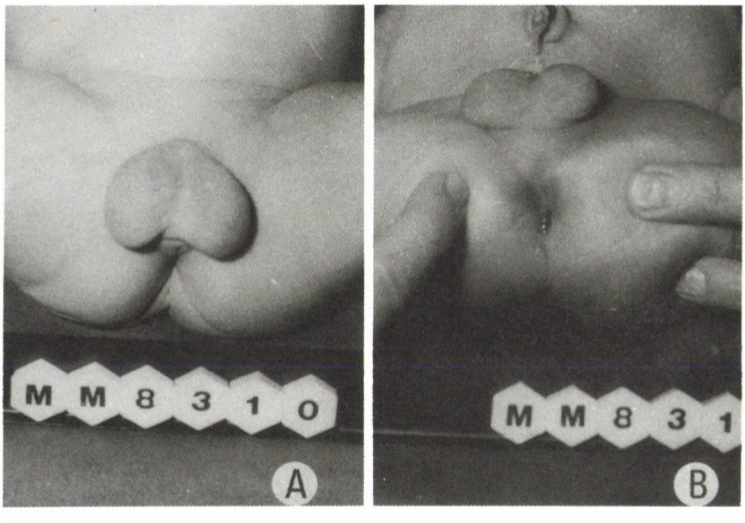

Fig. 4. Penile agenesis
a Absence of penis
b No external urethral oorifice on the perineum

CONCLUSIONS

Gender assignment is a neonatal surgical emergency and must not be delayed until the child is older. Change of gender after two years results in social disaster. Management of patients with ambiguous genitalia, which begins in the newborn period and sometimes extends through adolescence, should be done as a team approach with paediatric surgeons/urologists, genetists, and paediatric endocrinologists. Time of reconstruction seems optimal in term of the adequate size of the anatomy for technical consideration at age of three years. Well described techniques give quite satisfactory functional results.

In determining the type of surgical reconstruction, existing anatomy and not genetic sex are of the most important consideration. It is quite feasible to fashion a perfectly functional set of female genitalia but almost impossible to make a functional male phallus.

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