Surgery for intersex

Sarah Creighton MD MRCOG

J R Soc Med 2001;**94**:218–220 SECTION OF OBSTETRICS & GYNAECOLOGY, 31 MAY 2000

The physical features determining the sex of an individual are the karyotype, the internal and external sexual organs, the gonads and the secondary sexual characteristics which appear at puberty. Intersex conditions occur when there is a defect in the normal process of sexual maturation that results in abnormalities in any of these features. The management of these conditions is in the midst of great change. Every aspect is currently under review including diagnostic techniques, timing and nature of treatment including surgery, and information given to the patients. The true incidence of most of these conditions is unknown and great secrecy still surrounds them.

PRESENTATION

Intersex conditions can present at any time during life. The child may be born with ambiguous genitalia. Young girls may present with inguinal hernias containing testes or with primary amenorrhoea. They may be diagnosed as a result of having an affected sibling or they may have their abnormality as part of a generalized or major anomaly such as cloacal extrophy.

In the past, intersex disorders were classified into three groups—male pseudohermaphrodite, which includes XY females such as women with complete androgen insensitivity syndrome; female pseudohermaphrodite, which includes virilized women such as those with congenital adrenal hyperplasia; and true hermaphrodite.

However, the term pseudohermaphrodite is confusing and not helpful in describing the condition. In addition patients find it offensive. It is customary now to use simply the name of the condition.

SEX OF REARING

The decision as to sex of rearing requires full discussion between the clinicians (usually including a paediatric endocrinologist and a paediatric surgeon) and the parents. Biochemical and genetic support is imperative and a clinical psychologist experienced in this area must be involved from the start.

The decision to rear an infant with an intersex condition as female may be straightforward or very complex. For example, in congenital adrenal hyperplasia the child will have a female karyotype and will be born with normal uterus and ovaries. She is potentially fertile and in western cultures the decision to rear as female is seldom disputed. In girls with complete androgen insensitivity syndrome the phenotype is entirely female. The diagnosis is often made in adolescence or adulthood and again there is little debate about female gender assignment despite a male karyotype and the presence of testes. But the decision is much more complex with conditions such as partial androgen insensitivity or 5α-reductase deficiency where a child with a male karyotype and testes has ambiguous genitalia at birth. This child may be assigned to a female role and undergo gonadectomy to render him an infertile female.

Clinicians aim to choose the gender that carries the best prognosis for reproductive and sexual function and for which the genitalia and physical appearance can be made to look most normal. It is thought this will ensure a stable gender identity. If surgery is required, it is performed as soon as possible and no later than 24 months.

GENDER ASSIGNMENT

Current guidelines for medical and psychosocial care of infants are based on work by Money *et al.*[1,2] in the 1950s and 1960s. Money's premise was that children are psychosexually neutral until the age of 2 years and that what is required for a stable 'normal' gender identity is unambiguous genitalia and unequivocal assurance from parents as to the chosen gender. Money illustrated his guidelines by a case of a normal male infant (one of male monozygotic twins) who sustained traumatic loss of his penis at the age of 7 months, was reassigned female and underwent gonadectomy and surgical reconstruction. The child apparently developed a female gender identity and was used to prove the importance of 'nurture over nature' in gender identity formation. However, subsequently Diamond[3] reported that at the age of 14 years this patient rejected a female identity and at the age of 25 years married a woman and adopted her children. Even where the situation has been thought non-controversial, such as congenital adrenal hyperplasia with a female karyotype, there have been reported cases of gender change late in life[4]. There is now recognition that development of a stable sexual identity is a complex process. There are no indicators in infancy about gender identity later or sexual orientation, whether or not a child has an intersex

Department of Obstetrics and Gynaecology, University College Hospital, London WC1E 6AU, UK

condition. Other factors may be important. Animal work on the effects of sex steroids on the fetal brain has shown that male and female behaviour patterns can be changed by treating the animal with different sex steroids[5]. There are also anatomical differences in the male and female brain.

LONG-TERM OUTCOME OF FEMINIZING GENITOPLASTY

Associated with the above concerns is an increasing body of evidence, anecdotal and from support groups, that results of genital surgery are poor. There are few good long-term follow-up data and the effects of poor-quality surgery are hard to separate from other factors affecting gender identity and sexuality. Most studies that do include long-term follow-up have scanty details on sexual function, describing it as 'adequate' or 'satisfactory' with little expansion upon these terms. In early series no major problems were reported, but it later emerged that repeat surgery was commonly required at puberty to facilitate intercourse[6,7].

Older techniques such as clitorectomy (amputation of the whole clitoris) are no longer performed. Subsequent procedures to preserve as much tissue as possible by burying the clitoris led to painful erections. A current aim in surgery is to preserve sensation; for example, in clitoral reduction most surgeons now try to preserve the glans and the neurovascular bundle[8]. However, adult patients continue to report pain, scarring and loss of sensation. Comparison with women who have not had surgery as children is difficult since most children with these conditions do undergo feminizing genitoplasty. Vaginoplasty is usually performed along with the initial clitoral reduction as part of the feminizing genitoplasty. This too often needs revision at puberty and current debate centres on the need for vaginoplasty at such an early stage.

VAGINOPLASTY FOR VAGINAL AGENESIS

If female sex of rearing is chosen, then at some point a vaginoplasty is required. The timing and nature of vaginoplasty depends on whether the vagina is required for menstruation or solely for intercourse. This will of course depend upon the patient's medical condition. Women with a uterus—for example, those with congenital adrenal hyperplasia—will require a vagina for menstrual flow, whereas XY females (excluding those with gonadal dysgenesis who will usually have a uterus and vagina) will require a vagina only for intercourse. In women with androgen insensitivity the vagina may be of normal length, shortened or completely absent.

If treatment is required for vaginal agenesis, vaginal dilators should be used first[9]. With motivation and support, 'normal sexual function' has been reported in up to 78% of patients[10]. However, if dilators are not successful or not appropriate, surgery is the next option. There are many possible procedures for vaginoplasy and careful selection is crucial to success. In most procedures a neovaginal space is created and lined. The commonest vaginal approach is to line the neovagina with a split skin graft (McIndoe-Reed procedure). Good results have been reported[11] but contracture is a major complication, difficult to overcome. Other tissues such as amnion and peritoneum have been used to line the neovagina with good short-term results but long-term functional data are lacking[12,13]. Good results are also reported when intestine is used to line the neovagina[14], though this requires major abdominal and perineal surgery. Contracture and dryness do not occur but persistent mucus discharge can be distressing.

Clearly, the procedures for vaginoplasty are major operations with well-recognized complications, and patients must be fully counselled on the risks before any decision on surgery. There is a strong case for deferring operations until adolescence or later, so that patients can make a clear and informed choice.

THE PATIENT'S VIEW

There is increasing evidence of patient dissatisfaction with outcome[5] and a sensible policy is use surgery sparingly. However, it is impossible to define who actually 'needs' a clitoral reduction or vaginoplasty in childhood. The Intersex Society of North America—a well regarded and effective patient support group—recommends that no surgery should be performed unless absolutely necessary for the physical health and comfort of an intersexual child. The society regards vaginoplasty and clitoral reduction as cosmetic surgery that should be deferred until the patient can consent. This means leaving even the most virilized female babies without surgery and goes against current practice in the UK.

Some patients propose an even more radical approach—that there should be no attempt to allocate an intersexual child to male or female gender but that a third gender should be recognized. It would be difficult, however, to reach a consensus on which patients should be allocated to this new gender and there is no knowing whether these children would be any happier.

CONCLUSION

The main goal for clinicians working with intersex patients is to facilitate successful psychosocial adjustment. Until lately, genital surgery has been seen as the mainstay of treatment but recent evidence suggests that this is not so. Adult patients are unhappy and feel mutilated and damaged by surgery performed on them as young children, however worthy the clinician's motives. Although the technology for investigation and diagnosis of these conditions is improving

rapidly with advances in biochemical and genetic testing, there are still few long-term data on the results of intervention. Clinicians working in this field must step back and review their practice. Surgery may not be necessary. We need much more information to allow clinicians and parents to make informed decisions, and for this purpose multicentre research on long-term outcomes is essential.

REFERENCES

1 Money J, Hampson JG, Hampson JL. Hermaphroditism: recommendations concerning assignment of sex, change of sex and psychologic management. *Bull Johns Hopk Hosp* 1955;**97**:284–300

2 Money J, Hampson JG, Hampson JL. Imprinting and the establishment of gender role. *Arch Neurol Psychiatry* 1957;**77**:333–6

3 Diamond M. Sexual identity, monozygotic twins reared in discordant sexual roles. *Arch Sex Behav* 1982;**11**:181–6

4 Meyer-Bahlburg HFL, Gruen RS, New MI. Gender change from female to male in classical congenital adrenal hyperplasia. *Horm Behav* 1996; **30**:319–32

5 Hines M, Goy RW. Estrogens before birth and development of sex-related reproductive traits in the female guinea pig. *Horm Behav* 1985; **19**:331–47

6 Premawardhana LDKE, Huches IA, Read GF, Scanlon MF. Longer term outcomes in females with congenital adrenal hyperplasia (CAH): the Cardiff experience. *Clin Endocrinol* 1997;**46**:327–32

7 Alizai NK, Thomas DFM, Lilford RJ, Batchelor GG, Johnson N. Feminising genitoplasty for congenital adrenal hyperplasia: what happens at puberty? *J Urol* 1999;**161**:1588–91

8 Gearhart JP, Burnett A, Ownes JH. Measurement of pudendal evoked potentials during feminising genitoplasty: technique and applications. *J Urol* 1995;**153**:486–7

9 Frank RT. The formation of an artificial vagina without operation. *Am J Obstet Gynecol* 1983;**35**:1053

10 Rock J, Reeves L, Retto H, Baramki T, Zacur H, Jones H. Success following vaginal creation for mullerian agenesis. *Fertil Steril* 1983; **39**:809–13

11 Cali RW, Pratt JH. Congenital absence of the vagina: long term results of vaginal reconstruction in 175 cases. *Am J Obstet Gynecol* 1968; **100**:752–63

12 Ashworth MF, Morton KE, Dewhurst J, Lilford RJ, Bates RG. Vaginoplasty using amnion. *Obstet Gynecol* 1986;**67**:443–6

13 Ward G, Panda S, Anwar K. Davydov's colpocliesis: vaginal construction using peritoneum. *Curr Obstet Gynaecol* 1998;**8**:224–6

14 Martinez-Mora M, Isnard R, Castellevi A, Lopez-Orte P. Neovagina in vaginal agenesis: surgical methods and long term results. *J Pediatr Surg* 1992;**27**:10–14