Ethics in intersex disorders

Jyotsna Kirtane

Intersex cases are rare, and even medical personnel may not fully understand the finer nuances, implications and complexities of such cases. The arrival of a newborn is a highly emotional event in our country, and gender plays a key role. The birth of a boy is greeted with great enthusiasm. Not only is the continuity of the family assured but old age provision for parents is made. The arrival of a girl is attended by less bonhomie, and is linked with the travails of finding a suitable match and dowry.

In the Indian scenario, assigning gender is not easy, because most patients with ambiguous genitalia may end up as females.

The problem is further complicated by the timing of presentation.

If noticed at birth, it is seen in the form of abnormal genitals (congenital adrenal hyperplasia, micro penis and streak gonads).

If noticed at puberty, the symptoms are amenorrhoea, unexplained virilism (androgen insensitivity, 5 alpha reductase deficiency).

Both need thorough a diagnostic work-up before assigning gender. The latter may have another difficulty in that the child may have been raised as a male, but eventually be assigned female sex.

Ethical issues in intersex disorders are as follows: assigning sex at birth; birth registration and naming ceremony; getting investigations carried out expeditiously; deciding on gender; timing of surgery; telling the parents the truth; risks of malignancy, and talking to the patient.

Can we assign sex at birth?

This may be easy in congenital adrenal hyperplasia which also happens to be the commonest type of intersex disorder. Only estimation of 17 alpha ketosteroids and a female karyotype are required. In other cases like micropenis, streak gonads, etc., a thorough diagnostic work-up is required. This will include karyotyping, sonography, laparoscopy, genitography/scopy, biopsy plus many sessions of talking to relatives. Assigning sex in such cases must wait.

Dr Jyotsna Kirtane, *department of paediatric surgery, Bombay Hospital, Mumbai*

Birth registration and naming

The law requires that registration of birth be made within 40 days of birth. This will mean naming the parents, the child and the gender. In such cases a medical certificate attached to the registration form, helps the parents to get more time and delay the registration.

Investigations required

Most investigations can be carried out in the first month of life. Some like laparoscopy may need time, especially if the patient has associated defects or is an anaesthesia risk. It is imperative that the anxiety of the parents not be unduly prolonged.

Other problems such as deciding on the sex of rearing, necessary investigations and the time of operations are discussed with the following conditions:

Female pseudohermaphrodites

These are genetic females (46xx) born with hyperthrophied clitoris leading to ambiguity of genitalia, the commonest cause being congenital adrenal hyperplasia (CAH) commonly caused by 2-OH enzyme deficiency.

Confirmation of diagnosis is quick with demonstration of elevated plasma 17 alpha hydroxyprogesterone, with female karyotype, especially if there is a similar history in the family.

Treatment is to be instituted early with steroid replacement.

Sex of rearing: These children must be reared up as girls, as they have an excellent fertility potential. In India one may meet with a lot of resistance from the parents, if there is severe degree of masculinisation of the genitalia, and if the baby is already being reared as a boy, more so if the previous siblings are all girls. It is important for the doctor to convince the parents about rearing the child as a girl, reassuring them that surgery can rectify the genital anomaly.

Timing of surgery: This does not pose any ethical problems. Reduction clitoroplasty (with vaginoplasty if necessary) can be carried out in infancy.

Male pseudohermaphrodites

These are genetic males (46 xy). Ethical aspects of this group of disorders can be quite taxing to the concerned doctor. It is considered under its respective subtypes.

Complete androgen insensitivity (AIS)

As the external genitalia are completely feminine such a baby is not brought to the doctor at birth. Later on, presentation may be for inguinal hernia, or for primary amenorrhoea at puberty.

Diagnosis: In a girl presenting with inguinal hernia with palpable gonad in the hernia one should suspect intersex condition. Family history of sterile maternal aunts would strengthen the suspicion, as would presence of scant axillary or pubic hair in the mother. A per rectal examination is then necessary to feel for the uterus or cervix. Karyotyping would confirm the diagnosis.

Management: These children are best reared as girls as they cannot be distinguished from normal girls at all. They develop breasts at puberty. The only surgery they require is gonadectomy and vaginoplasty. The timing of gonadectomy is controversial. We prefer to defer this till after puberty as this allows for natural female development to take place at puberty as a result of conversion of endogenous testosterone to oestradiol. In the West, most surgeons and parents prefer to

undertake surgery early with hormone replacement therapy.

Telling the parents: How much should one tell the parents? The statement that their daughter is actually a "boy" may shock them. If the parents are intelligent and educated, capable of understanding, one should explain the genesis of the condition. The most important thing is to reassure them that their child will grow up as a "normal" girl except that she will not menstruate and will not reproduce. She can be sexually functional with a vaginoplasty. Also, she will have to undergo surgery after puberty to prevent malignancy in her gonads.

Incomplete androgen insensitivity and 5 alpha reductase deficiency:

These children are born with incomplete masculinisation of the genitals. Even so, some of these are reared up as boys or brought to the doctor later on for hypospadias. Diagnosis may take time as the necessary investigations are complex, if one has to differentiate between incomplete AIS and 5 alpha reductase deficiency. It is necessary to differentiate between the two as the latter get masculinised after puberty whereas the former develop female secondary sexual characters. At times, the investigations may not be available at all. One has to then take a decision based on size of phallus, parental wishes and the response to depot preparation of testosterone injections.

There is no controversy in cases the phallus is small and response to testosterone is poor, in alloting a female sex of rearing, with gonadectomy, feminising genitoplasty and hormone therapy.

The parents must be told the entire truth, that their child is genetically a boy, but would function better as a girl. If the child has to reared as a boy, a urethroplasty is necessary and can be carried out around one and a half years of age. We prefer to do it in one stage, using either Thiersch's duplex or a buccal mucosa graft.

In cases of feminising genitoplasty, reduction clitoroplasty with creation of labia minora can be done in infancy, deferring the vaginoplasty to a later date. The two procedures may also be combined at around one and a half years of age.

True hermaphrodites and mixed gonadal dysgenesis

These children generally present with ambiguous genitalia and arriving at a diagnosis usually takes time, as gonadal biopsies with or without laparotomy/laparoscopy and karyotying is required before conclusively proving the diagnosis.

This delay can be very trying for the parents. It is necessary for the doctors to give the parents, some idea regarding the different possibilies. They need to be told, that regardless of the outcome of the investigations, their child will be managed in an adequate fashion.

Sex of rearing: This is dictated by the phenotype. Children having adequate sized phallus are reared up as boys. Surgery includes, besides repair of hypospadias, excision of all ovarian tissue and female internal genitalia and orchiopexy if the testes are not adequately descended and fixed. Similarly in children with small phallus, a feminising genitoplasty is done, excising all testicular tissue.

The timing of surgery depends upon the age at which the child is brought to the surgeon. Ideally all surgery should be over before the child starts schooling.

In cases of mixed gonadal genesis, the same approach is used, excising the streak gonad.

Fertility potential: Having overcome their initial period of anxiety and accepted the sex of rearing of their child, the parents are anxious to know the details of future prospects of the child – regarding sexual function and fertility potential. Fertility is unlikely in many intersex conditions, with the exception of CAH. Some true hermaphrodities are also known to have been fertile, but it is not wise to predict the fertility potential in other cases, except CAH.

Cautioning parents on gonadal malignancy

Children with intersex disorders and/or with abnormal sex chromosomes are at an increased risk for development of malignancy, particularly in their gonads. It is seen more commonly in: complete AIS, with malignant germ cell tumours, usually in adult life; all gonadal dysgenetic disorders in patients having a Y chromosome, who may develop gonadblastomas even in their first decade, and mixed gonadal dysgenesis where Wilm's tumour of the kidney has a higher incidence.

It is of paramount importance to alert the parents of the possibility of malignancy, occurring in later life, and hence to maintain a follow-up record.

How much should the child know?

Once the child grows up, he or she may have several disturbing queries regarding secondary sexual characters, amenorrohoea, etc. It is best not to disclose to the patients the true nature of the underlying intersex disorder. This may shatter adolescent patients completely, and perhaps even induce suicidal tendencies.

However, the deficiencies or abnormalities can be explained to the parents, who in turn may talk to the patient with or without the help of a psychologist.

Genetic counselling: This is easy in a case where the mode of transmission within the family is known, such as adrenogenital syndrome, an autosomal recessive disorder, in which the chance of giving birth to an affected child is 1:4 and one with sexual ambiguity is 1:8 (only the female child will have ambiguity).

Conclusion

It is clear that tbe ethical issues in managing intersex disorders are very complex. Even after waiting for months to announce the birth of a child to relatives, parents are unclear if it is a girl or a boy. They are disturbed by the fact that if it is a boy his sexuality may be inadequate and if it's a girl she may not menstruate or procreate. Even worse is explaining all this to the child. In the Indian context this can be truly devastating.

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