IDENTICAL INTERSEXUAL DISORDER IN TWO SIBLINGS

by

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Normal sex differentiation and function is important and vital to the survival of human race. Individuals who have gross sex differentiation anomalies cannot fit themselves properly in the society unless the condition is diagnosed at a very early age and corrective surgery carried out, where necessary.

Early testis of the foetus in utero has a twofold function—

(i) To provide the necessary stimulus for normal male development.
(ii) To inhibit the growth of Mullerian structures.

If the function of the testis is partially suppressed in utero the individual is born with various degrees of sex anomalies, though possessing a XY genotype. We record here two identical cases of intersex in two siblings where the anomalous external genitalia present at birth were ignored and no investigation or treatment carried out in childhood.

Case 1

B.G., aged 19 years, presented at Nilratan Sircar Hospital on 1-3-72 with complaints of primary amenorrhoea and hoarseness of voice for the last 6 years.

At the age of 14 years she attended some other hospital with complaints of primary amenorrhoea, non-development of breasts and hoarseness of voice.

On examination 5 years previously the patient was found to have male physical features and slight excess growth of hair. Breasts poorly developed. Phallus enlarged with hypospadias. Blind and narrow vagina; vulva not feminine but looked like bifid scrotum. Gonads not felt in the vulva.

Chromatin negative buccal smear. Pelvic pneumogram showed absence of uterus and gonads.

Hormone excretion patterns on 26-10-67: 17-Ketosteroid 7.8 mg. and 17-hydroxycorticosteroids 10.9 mg. in 24 hours urine. Oestrone in 24 hours urine—5.4 μg. Oestradiol in 24 hours urine—1.5 μg.

As the patient had been reared up as a girl and was attending a girls' school, it was decided to make her as feminine as possible with corrective surgery.

At operation the phallus was removed, maintaining the normal anatomical relationship of phrenum and prepuce. The gonads were palpated in the inguinal canal above the external inguinal ring. By a transverse incision both inguinal canals were opened. Gonads were exposed and found to have the appearance of testis. The gonads were removed. Epididymis was very rudimentary and there was no vas deferens. Abdomen was opened by the same transverse incision. There was a central transverse ridge of mesodermal tissue representing the uterus. There was no prostate fallopian tubes or ovaries. Kidneys were in normal position and suprarenals were of normal size.

On 1-3-72 the patient had a height of 5'-7½", weight 55 kgs. There was a fairly good breast development as a result of oestrogenic hormones received by her after previous operation (Fig. 1).

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Case 2

S.C., aged 16 years, younger sibling of Case 1 presented at the endocrine clinic of N.R.S. Hospital on 1-3-72 with complaints of primary amenorrhoea, excess growth of hair, and appearance of a swelling in right groin during exercise, associated with pain and tenderness over the swelling. The swelling and pain disappeared on rest.

Family History: She has 3 normal brothers. Elder "sister" as described above. Younger sister aged 10 years, not started menstruation as yet. No history of sex anomaly or amenorrhoea among father's or mother's sisters.

On Examination:

Height 5'-6", weight 58 kgs. Masculine features. Male distribution of hair. Breasts poorly developed (Figs. 2 and 3). Phallus about 1" in length (Fig. 4).

Inguinal gonads. Inguinal hernia on right side and the right gonad could be brought down to the right labium majus. Labia looked more like bifid scrotum (Fig. 5). Narrow and blind vagina, admitting one finger; length 2½". Uterus could not be felt by vaginal or rectal examination. Buccal smear—Chromatin negative

This patient also was attending a girls' school; so it was decided to remove the testes and make her as feminine as possible by plastic surgery undertaken on 20-6-72. Both the inguinal canals were opened by a transverse incision. The gonads had the appearance of testes (Fig. 6). Both the gonads were removed. The abdomen was opened through the same transverse incision. No Mullerian remnant was seen and there was no prostate. Epididymis was very rudimentary and there was no vas deferens. During closure of the abdomen both the inguinal canals were repaired. The phallus was not removed as it was thought that the size would diminish after removal of the testes. Similarly, no redundant skin was removed from the right labium majus.

Histological examination of the gonads showed the structure of testis (Fig. 7). Examination of the patient on 14-8-72 showed the phallus to be smaller in size.

Examination on 30-10-72, 2-2-73 and 15-8-73 showed that phallus was much smaller in size and the hirsutism was much less marked. Patient herself thought that there were less hair.

The youngest sister of the patient, aged 10 was examined. She had no sex anomaly. Uterus was normal in size for this age. Breasts fairly developed (Fig. 8). Buccal smear—Chromatin positive.

Discussion

Diagnosis and correction of anomalous sex should be undertaken in early childhood to avoid embarrassment to the family and the patients concerned. If these two cases had presented in childhood within a few months of birth, proper guidance could have been given to the parents as to the sex of rearing of the children. The general guideline is that one should take into consideration the wish of the parents as well as the end results which will be found after corrective surgery. Of course, the parents should be made to realise that only partial sexual and no reproductive function is possible in the type of cases described here. In these particular cases if it was decided that the sex of rearing would be female, the corrective surgery should have been performed much before puberty.

The intersexual disorders are very often genetically transmitted as was found in the two siblings here. We have two other siblings aged 2 years and 5 years, almost having the same type of intersexual disorders; they are having plastic surgery. Three cases of testicular feminisation syndrome in the same family have already been published by the authors (Gun and Co-workers, 1973). The defect of the two siblings mentioned here is genetically transmitted through a sex chromosomal or autosomal recessive gene (Milner *et al*, 1966), although the exact metabolic defect is not clear.

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See Figs. on Art Paper II-III