PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS (P.P.S.H.)

by

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Nowakowski and Lenz provided the first clinical and genetic definition of this syndrome in 1961. A recent extensive review of the literature has aided in delineating this condition as a well-defined clinical and genetic entity.

PPSH, a condition of male pseudohermaphroditism with ambigous external genitalia is characterized by a phallus of intermediate size with a ventral urethral groove and a perineal urethral meatus. There is a blindly ending vagina or urogenital sinus which may open either into the urethra or the perineum. A labia majora like cleft scrotum frequently contains normally developed testes. Wolffian duct derivatives show normal male differentiation; Mullerian duct structures are absent. The phallus is usually mistaken for an enlarged clitoris and a perineal opening for a vagina. Thus, these persons are frequently reared as girls. Extragenital malformations are not known in this disorder and affected persons are of normal intelligence. Masculinisation at puberty results in masculine body build, axillary, fascial and pubic hair and deepening of the voice. Breast development is absent (Sarto 1972).

Urinary gonadotropin levels are normal. Plasma testosterone and urinary 17 ketosteroid levels are in normal range for males (Qultz *et al*, 1972). Plasma testosterone concentration in the testicular vein is much higher than in peripheral veins. Thus, testicular function is initially completely normal. Histologic examination of the testes may show tubular degeneration with hyalinization and clumping of interstitial cells (Opitz *et al* 1972). We had one such rare case which is presented.

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CASE REPORT

G. S. aged 20 years, a young unmarried girl of average intelligence was admitted in Govt. Medical College on 27-2-1979 with complaint of not having attained menarche.

Family History

The patient being the last child of the family 2 brothers normal, sister is elder to her, married, has 3 children; no history of consanguinity of parents.

On general examination average build, breasts not developed with poor nipple and areola. Axillary hair normal. Pubic hair of masculine distribution. Other systemic examination normal. No other congenital abnormalities noted.

External Genitalia

A small penis of 1½ cm size. Testes 1½ cm size in both scrotal sacs. Testicular sensation present. Urogenital sinus present at the perineum and also ventral urethral groove. No separate vaginal orifice. Spermatic cord felt and normally differentiated. Mullerian duct structures are absent as made out by rectal examination. (Fig. 1 and Fig. 2).

Investigations

HB% 12 gm%, routine urine and haemogram within normal limit. I.V.P., X-ray chest and X-ray skull revealed nothing abnormal. 17 Ketosteroid 6 mg/24 hours, Sex chromatin Negative for Bàrr bodies.

Testicular Biopsy

Shows seminiferous tubules with incomplete spermatogenesis, thickening of basement membrane. Interstitial cell hyperplasia is present.

Management

Laparotomy was done on 16-3-79. Mullerian duct structure not developed. Bilateral gonodectomy was done after ligating spermatic cord. As this patient was reared as a female gonadectomy was done and patient kept on estrogen, progesterone therapy.

Discussion

This disorder is thought to be due to a (Temporary ?) partial insensitivity of external genitalia and urogenital sinus to androgens. The functions of the foetal testis with respect to the induction of Wolffian duct differentiation and inhibition of Mullerian duct structure appear normal (Sarto, 1972; Short, 1967). Genetically this condition may be caused by homozygous state of a rare autosomal recessive mutation limited in expression to males (Sarto, 1972). PPSH persons reared as males may require urologic surgery to correct hypospadias and to remove the vaginal pouch. Whether these males are capable of reproducing is not known at present and those when reared as females and assigned females should have pre-puberal gonadectomy and estrogen and progesterone therapy to stimulate the development of female secondary sex features. This condition may be difficult to differentiate from incomplete testicular feminisation syndrome.

Summery

The case of PPSH syndrome, a genetic disorder, is reported. When born, these babies are reared as female by some parents and as males by others. The difficulty in assignment of sex of such individuals and pathogenesis and genetics of such cases are discussed.

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See Figs on Art Paper VII-VIII