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

by

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Pseudoperineo-scrotal hypospadias 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 the 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 result in masculine body build, axillary, fascial and pubic hair and deepening of the voice. Breast development is absent. (Sarto *et al*, 1972).

Urinary gonadotropin levels are normal. Plasma testosterone and urinary 17-ketosteroid levels are in normal range for males. (Opitz *et al*, 1972) Plasma testosterone concentration in the testicular vein is much higher than in the peripheral veins. Thus, testicular function is initially completely normal. Histologic examination of the testes may show tubular degeneration with hyalinisation and clumping of interstitial cells. (Opitz *et al*, 1972). We had two such rare cases for investigation for cytogenetic study and genetic counselling at the human genetics section, Pathology Department. There is no Indian literature available on these rare cases hence they are presented in this paper.

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CASE REPORTS

Case 1.

S.B. aged 25 years, a young man of average intelligence, phenotype male, tailor by profession came for investigation for genetic sex and for marriage counselling. He had desire to marry a girl. He was given by a quack a course of testoviron injections but there was no improvement except that he had seminal emissions as per the history. Consanguinity of parents present (Uncle niece). (Fig. 1).

External Genitalia: A small penis of 1 cm size. Testes 1 cm size in both the scrotal sacs. Scrotal skin rugose, testicular sensation present. Urogenital sinus present at the perineum and also ventral urethral groove. No separate vaginal orifice, bifid left scrotum, Wolffian duct derivatives like spermatic cord felt and normally differentiated. Mullerian duct structures are absent as made out by rectal examination. Prostate normal. (Fig. 2 and fig. 3).

Pubic hair of masculine distribution. Axillary hair normal. History of passing occasionaly white seminal fluid per urethral opening. Other systems—nil abnormal. No other congenital anomalies noted.

Investigations

HB: 10 Gms%, 17-Ketosteroids 5.5 mg/24 hours.

Thin prostatic fluid: Showed no spermatozoa. In urine, few epithelial cells seen.

Testicular Biopsy: Sclerosis of some tubules

and few tubules are immature and show thickening of B.M., no spermatogenesis noted in the tubules. Leydig cell clumps in the interstitial areas suggesting hyperplasia.

Sex chromatin: Barr bodies negative.

I.V.P. No Renal, Ureteric anormalies noted.

Karyotype analysis: 46/XY. Genetic male.

Case 2.

Baby H.B.S. 5 weeks old with ambiguous external genitalia referred from the Naval Hospital, for genetic counselling to know the genetic sex.

Family History: This is the third sibling of the family. Consanguinity of the parents present. The two elders were sisters and no abnormality in any of them. External genitalia show bifid scrotum, a urogenital sinus in the perineum with urethral groove running behind the penis which is slightly adherent at the base with skin. Penis of 1 cm size suggesting pseudovaginal perineoscrotal hypospadias (Fig. 4). Sex chromatin negative for Barr bodies. Genetic sex male, karyotype analysis on 16-10-73: 46/XY. Parents were advised to rear the child as male and surgery to be done later to correct hyposapadias before 10th year.

Discussion

This disorder is thought to be due to a (Temporary?) partial insensitivity of the 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), as noted by the consanguinity in the parents of this patient. 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 female should have prepuberal gonadectomy and oestrogen and progesterone therapy to stimulate the development of female secondary sex features. This condition may be difficult to differentiate from incomplete testicular feminisation syndrome. Chromosome analysis excludes mixed gonadal dysgenesis and is an important investigation necessary to determine genetic sex.

Summary

Two rare cases of PPSH syndrome, a genetic disorder are 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 is discussed. Pathogenesis and genetics of such cases were discussed.

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References

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See Figs. on Art Paper VIII