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RESEARCH ARTICLE

SEVERE HYPOSPADIAS WITH APHALLIA - CASE OF CONFUSED SEX

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ABSTRACT

Hypospadias is a birth defect of the urethra in the male that involves an abnormally placed urethral meatus .Instead of opening at the tip of the glans penis , a hypospadiac urethra opens anywhere along a line running from the tip along the ventral aspect of the shaft to the junction of the penis and scrotum or perineum. Aphallia is the penile agenesis is a very rare genitourinary anomaly that has profound surgical and psychosocial implications. Penile agenesis cases have 46,XY karyotype .Early assignment of female gender and feminizing reconstruction of the perineum is the goal treatment in cases of Aphallia. Disorders such as severe hypospadias presenting ambiguous genitalia have serious and potentially life long consequences for affected individuals and, depending on the underlying cause ,are likely to entail surgery in childhood and in later life ,psychosocial and psychosexual support and possible fertility treatment including assisted conception. Genetic males with severe hypospadias who will wrongly bring up as females has to be reassigned as males .These disorders results partial or complete failure of the fusion of urethral groove and failure of the development of genital tubercle.

INTRODUCTION

Hypospadias are among the most common birth defects of male genitalia , but widely varying incidences have been reported from different countries ,from as low as 1 in 4000 to as high as 1 in 125 boys (Stephens *et al.,* 1996).There has been some evidence that the incidence of hypospadias around the world has been increasing in recent decades. Most hypospadias are sporadic, without inheritance or family recurrence. For most cases, no cause can be identified though a number of hypothesis related to inadequate androgen effect, or environmental agents interfering with androgen effect, have been offered. Prenatal testosterone, converted in the genital skin to dihydrotestosterone, causes migration of skin fibroblast to fully enclose the urethral groove in fetal males, normally resulting in an enclose urethral groove in fetal males, normally resulting in an enclosed penile urethra by second trimester of pregnancy (Baskin, 2000). Failure of adequate prenatal androgen effect is therefore thought to be involved in many cases, making hypospadias a very mild form of intersex. In a minority of cases a postnatal deficiency of or reduced sensitivity to androgens can be demonstrated. These are often associated with a chordee, and in severe cases a residual perineal urogenital opening and small phallus. This combination of birth defects is referred to as pseudovaginal perineoscrotal hypospadias and is part of spectrum of ambiguous genitalia (Hym and Kolon, 2004).

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MATERIALS AND METHODS

During the fourth to seventh week of development the external genitalia are sexually undifferentiated. Early in the fourth week proliferating mesenchyme produces a genital tubercle in both sexes at the cranial end of the cloacal membrane. The genital tubercle soon elongates to form phallus. Labioscrotal swellings and urogenital folds soon develop on each side of the urogenital membrane. Musculanization of the indifferent external genitalia and mesonephric ducts are caused by testosterone produced by interstitial cells of leydig of fetal testis by about 8th week. Testosterone production is stimulated by hcG, which reaches its peak at 8th to 12th week period. Mullerian inhibiting factors, produced by sertoli cells of foetal testis, and suppress development of the paramesonephric (mullerian) ducts. As the phallus enlarges and elongates to become the penis, urogenital folds, lining the lateral walls of the urethral groove on the ventral surface of the penis, fuse in the midline to form the spongy urethra. Line of fusion is the penile raphe. *Corpora cavernosa* and *Corpora spongiosum* develop from mesenchyme in the phallus. Labioscrotal

swellings of each side fuse to form the scrotum and the time of fusion of these folds in the midline form the scrotal raphe.

RESULTS

A term male baby born out of non- consangnious marriage delivered normally in the labour room of Hospital weighing 2.8 kg was born to 28 year old primi-mother. There was no history of congenital anomaly in the family. There was no history of any significant drug intake or radiation exposure during pregnancy. Antenatal and perinatal period was also uneventful. On examination it was found there was complete absence of penis, but scrotum was well developed with both testes. On examination a small orifice was found at perineal area, at the anal verge. With these two observations we have noticed that the baby is having severe hypospadias and Aphallia (Figure 1). Baby had no other congenital anomalies, ultrasonography of whole abdomen was within normal limit. Karyotype was not done as the parents did not allow to do further investigation.

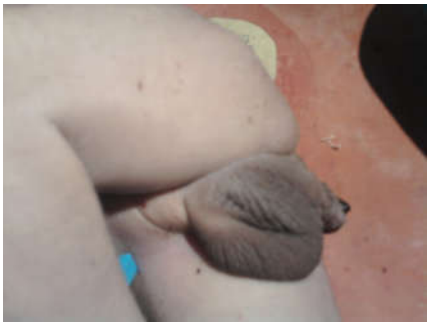

Fig. 1. Aphallia with hypospadias

DISCUSSION

Hypospadias may be an isolated defect or a phenotypical component of a more complex condition such as an intersex state. Hypospadias ,in boys ,is defined as an association of three anomalies of the penis: an abnormal ventral opening of the urethral meatus that may be located anywhere from the ventral aspect of the glans penis to the perineum, an abnormal ventral curvature of the penis called chordee, and an abnormal distribution of fore skin with "hood" present dorsally and deficient foreskin ventrally (Baskin *et al.,* 2004). The second and third characteristics are not present in all cases. Hypospadias is classified into distal, middle, and posterior. The urethral meatus opens on the glans penis is categorized as first degree hypospadias (Skoog *et al.,* 1989). When the urethra opens on the shaft is second degree hypospadias, when the urethra opens on the perineum is third degree of hypospadias. Ahallia is classified in to three types post sphincteric meatus located on a peculiar appendage at the anal verge, pre sphincteric prostatorectal fistulas and urethral atresia (Tiwari, 2003). First case of aphallia was reported by Imminger in 1853. Aphallia is diagnosed by the absence of corpora cavernosa and corpora spongiosum along with external urethral opening situated in the midline of perineum. Intersexuality represents a rare but important group of disorders, which usually present at birth with ambiguity of the external genitalia. It is imperative that these conditions are recognized early and steps taken to identify the underlying causes as in some cases, a delay may result sudden collapse and death from an underlying metabolic disorder. When the phenotypic appearance of the genitalia is ambiguous, appropriate biochemical, radiographic, and chromosomal studies should be completed (Reiner *et al.,* 2000). The degree of external virilization should be carefully noted and recorded, careful photographs recorded and parents need to be advised that a sex assignment decision will be based on the appropriate data. Gender assignment /reassignment and surgery should be deferred until such an age when the individual can make informed decisions. Genital reconstruction in the form of hypospadias repair, psychosocial changes and psychosexual counseling will help in managing these children. Assignment of genetic males to the female sex because of severe hypospadias and phallic inadequency can result in unpredictable sexual identification. Androgens influence prenatal brain development as well as postpubertal activity, interests and libido. Children who are genetic males with severe hypospadias but wrongly bringing up as females at birth should be reassigned as males. Psychosocial changes in environment, school, home and relations must be made. Psychosexual counseling and orientation will help the children to cope with their new identity.

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