Case Report

Penile Agenesis Associated with Anorectal Malformation and Bilateral Renal Agenesis: A Case Report and a Review of the Literature

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Abstract

Penile agenesis is complete absence of the penis in an otherwise normal phenotypic and genotypic male at birth that results from failure of development of the genital tubercle. It is an extremely rare anomaly that may be associated with anomalies in other organ systems, the extent and severity of which may affect the prognosis. The management is challenging and may have far reaching implications for the individual and family. While gender reassignment with bilateral orchidectomy and feminising genitoplasty has been carried out for most patients, significant psychosexual and social issues related to the male identity may occur due to foetal or postnatal sex steroid imprinting. We report a neonate with penile agenesis with bilateral renal agenesis and anorectal malformation.

Keywords: Anorectal malformation, penile agenesis, renal agenesis

Introduction

Penile agenesis was first reported by Barthélemy Saviard, a French surgeon, in 1701, in an article he captioned as 'child who had no rod'. However, a more elaborate description was by Imminger in 1853.[1] Penile agenesis, or aphallia, is due to the absence or failure of the development of the genital tubercle.[2,3] Rarely, rudimentary erectile tissue may be present.[2] The karyotype is almost always 46, XY.[2] It may be isolated or associated with other anomalies and variable degrees of connection with the gastrointestinal tract.[3] The reported incidence is one in ten million to thirty million,[2] and appears to be rising. When associated with other anomalies the incidence of stillbirth or early neonatal death is about one in three cases.[3]

Case Report

The patient was a 5-h-old neonate noted to have an absence of the penis and anus at birth. Mother had antenatal care in a peripheral centre and the pregnancy was uneventful. Obstetric scans were said to show no anomaly other than inability to ascertain the sex of the foetus. He was low birth weight (1.9 kg); however, the gestational age at birth could not be ascertained from the mother, the right testicle was intrascrotal, whereas the left was intracanalicular. There was no opening on the perineum and the gluteal fold was poorly developed [Figures 1and 2].

He had an abdominal ultrasound scan which revealed bilateral rudimentary reniform structures measuring 2.2 cm × 0.5 cm and 1.8 cm × 0.5 cm, right and left, respectively, with minimal vascularity noted on Doppler interrogation. Urinary bladder could not be visualised. Serum electrolyte, urea and creatinine panel and packed cell volume were all within normal limits.

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Figure 1: Absent anal opening



Figure 2: Absent penis with a well developed scrotum

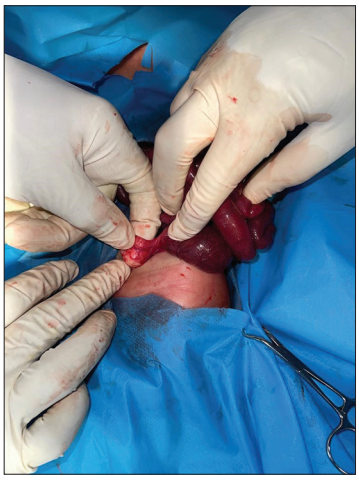


Figure 3: Blind-ending sigmoid colon with fibrous connection to an atretic bladder



Figure 4: Divided colostomy

Management was multidisciplinary involving paediatric surgeons and paediatric nephrologists. The paediatric nephrologist recommended that the child be placed on 10% dextrose water. Parents were counselled on the need for faecal diversion and possible urinary diversion. Intraoperative findings included a distended blind-ending sigmoid colon connected to a fibrous band in the pelvis which is likely an atretic urinary bladder [Figure 3]. The ureters were absent bilaterally and no Müllerian structures were seen. He had a defunctioning colostomy [Figure 4]; however, urinary diversion was not possible due to the absent ureters and atretic bladder. The patient was placed on maintenance 10% dextrose water and commenced breastfeeding on the second day after surgery. Serum electrolytes, urea and creatinine result done on the second day after surgery was within normal limits. However, on the fourth day of life, he was noticed to have generalised body swelling and he died shortly thereafter. Autopsy could not be done as the parents declined to give consent.

Discussion

The congenital absence of a penis in a biological male poses a major challenge in its management. The scrotum is normally developed and the testes are usually normal.[3,4] The urethra when present ends in the perineum or in the anterior wall of the rectum. The genital tubercle is the primordium of the penis and appears during the fourth week of intrauterine life.[5] Normal penile development requires an intact hypothalamic–pituitary–testicular axis. Although Skoog and Belman in 1989 proposed an anatomical classification of patients with penile agenesis based on the relationship of the ectopic urethral meatus to the anal sphincter,[6] the classification developed by Evans *et al*.[7] is prognostically significant and is based on the presence or absence of major associated anomalies. Evans categorised patients with aphallia into three groups. The first group is associated with renal aplasia, renal dysplasia and other caudal abnormalities. Our patient belongs to this group with bilateral renal agenesis and anorectal malformation. Sixteen percent of patients with penile agenesis belong to this group and mortality is high. The second group has fewer associated malformations and lower mortality rate, whereas the third group is not associated with any specific pattern of anomalies.[7]

The treatment of penile agenesis appears controversial,[8] further compounded by its extreme rarity which results

in a dearth of global experience in penile reconstruction in these patients. This appears to be the major reason that Western countries tend to assign female gender to infants born with aphallia.[9] Gender reassignment involves bilateral orchidectomy and feminising genitoplasty in the neonatal period and construction of a neovagina later in life.[3] Male gender assignment and early penile reconstruction might avoid the frequent sexual identity problems that may inevitably progress with age due to prenatal imprinting of testosterone on the brain. Prenatal and early postnatal androgen exposure have been shown to contribute significantly to male psychosexual development and identity.[9] Hence, the need for adoption of a multidisciplinary approach with full evaluation by an expert ambiguous genitalia assessment team alongside with parental counselling.[2] With current advances in penile reconstruction and urethroplasty, sexual and voiding function may possible, although reproductive function is unknown.[3]

Conclusion

Isolated penile agenesis is associated with profound psychological distress and poses significant challenges in its management. However, when associated with bilateral renal agenesis as in this index patient, it becomes a lethal variation of an extremely rare disorder.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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