AGENESIS OF PENIS: REPORT OF TWO CASES

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Abstract - In this report we introduce two cases of penile agenesis. The first case was a newborn with male appearance and a well developed scrotum containing two normal testes, but the penis and urethral meatus were absent. The bladder was connected to the rectum by an intra-abdominal canal. The second case was a premature newbron of 32 weeks, who died in the first day of life. In addition to penile agenesis, he had undescended testes, imperforate anus, duodenal atresia, annular pancreas, kidney agensis, and toe anomalies.
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Key words: Agensis of penis, aphallia, congenital malformation

INTRODUCTION

Agenesis of penis is a rare congenital malformation. Its incidence is approximately one in 10 million to 30 million (1, 2). Because of associated life-threatening congenital malformations the fetus dies in the uterus in one third of cases. Only 75 cases have been reported in the world literature until 1995 (3, 4). We report the first published cases of aphallia in Iran. Location of the meatal orifice is an important prognostic factor in this malformation. The more proximal the urethra ends, the greater is the likelihood of having associated lethal anomalies and the more probable is intrauterine death of the fetus (5). The infant's gender must be changed to female as soon as possible by gonadectomy to prevent future psychosocial problems (6, 7).

Case 1

This two-day old newborn of 34 weeks gestational age was admitted on September 3, 1993. The infant had moderate abdominal distension, vomiting, no meconia defecation and no urination. The mother was 26 years old with a history of abortions at 2nd and 3rd trimester. The father was healthy and had six normal children from his first wife. There was no consanguinity of the parents. The mother had taken chlordiazepoxide, aspirin, ampicillin, folic acid, ferrous sulphate and multivitamin preparations during pregnancy. Birth weight was 2900 grams, length 48 centimeters and head circumference 34 centimeters. The newborn had fever, respiratory distress, and abdominal distension. There was no penile structure visible at its normal location. The scrotum was normal and contained two normal testes (Fig. 1). The anus was situated at its normal position. After dilation and irrigation of the rectum, voluminous meconium was evacuated, abdominal distension subsided and respiratory distress vanished. The bladder was distended and palpable near the umbilical region. This was evacuated by suprapubic puncture. Abdominal X-ray showed many air-fluid levels. In ultrasonography, both kidneys were hydronephrotic. Routine tests were normal. Blood urea nitrogen (BUN) was 54 mg/100 ml, phosphorus was 3.2 mg/100 ml. Emergency catheter cystostomy was performed. Retrograde cystography showed an abnormal connection between bladder and rectum as well as vesicourinary reflux (Fig. 2.). Buccal smear was chromatin negative and the aryotype was 46XY. Isotope scanning of the kidneys showed near normal function with moderate hydronephrosis and megaureter. After explanation of the newborn's condition to his parents, bilateral orchiectomy was done at his 12th day of life and he was discharged with a cystostomy.

Case 2

This newborn was born at 32 weeks gestational age on January 7, 1996 to a 35-year old healthy mother. Birth weight was 1700 grams. The parents were not consanguinous. The infant had Potter's face. The penis was completely absent and the scrotum was devoid of testes. In addition to the

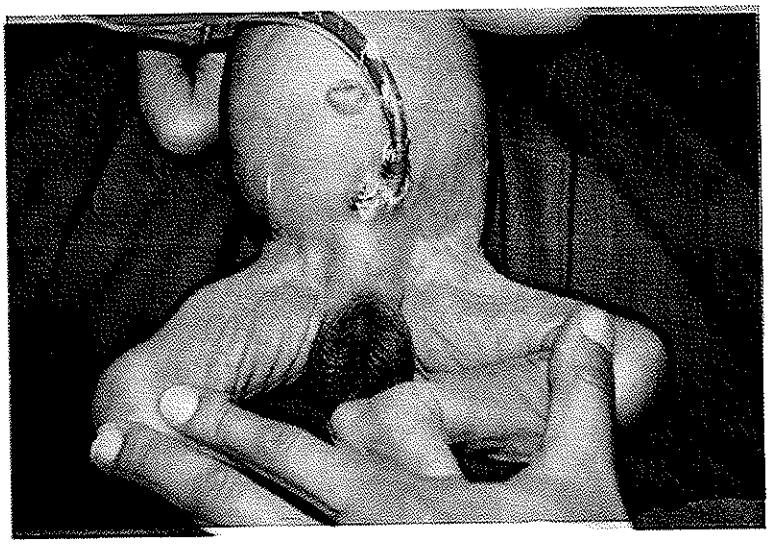

Fig. 1. Case 1. No penile structure visible at its normal location. Normal scrotum and two normal testes

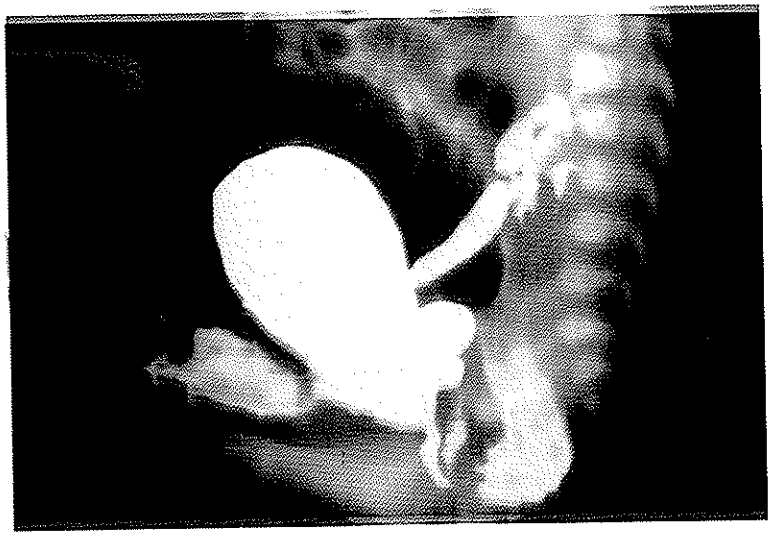

Fig. 2. Case 1. Retrograde cystography

clinical findings, a duodenal atresia, annular pancreas, agenesis of the left kidney, were found.

DISCUSSION

Penile agenesis or aphallia is a very rare congenital malformation. Its etioligy is unknown. Developmental deficiency of the genital tubercle in embryonal period may by the cause (9, 10). Development of the male genital system occurs under influence of fetal testicular androgens so that elongation of the male genital system occurs under influence of fetal testicular androgens. The elongation of the genital tubercle by any cause may lead to penile agenesis, then the urogenital sinus descends towards the genital tubercle, so urethral meatus opens in the perianal area to the rectum (7, 11). The associated anomalies consist of renal ectopia, renal hypoplasia, undescended testis, vesico-ureteral reflux, prostatic agenesis, lung hypoplasia, absence of median raphe, imperforate anus, rectal agenesis, rectal stenosis, rectovaginal fistula, inguinal hernia, sacral agenesis and hemivertebrae (8 - 17).

About one-third of the patients die intrauterine because of severe concurrent respiratory or genitourinary anomalies. Usually the karyotype of the patients with penile agenesis is 46XY. In addition to emergency cystostomy for improvement of renal function and urination, many other surgical procedures are required for gender assignment and reconstruction of anatomic disorders. Penile construction is often unsatisfactory. Orchiectomy, urethal transposition and labial construction are required. Vaginoplasty should be deferred until a functional vagina is necessary. Orchiectomy is usually done on the 10th to 60th day of life (6 - 7). At puberty, hormone therapy (estrogens) is neccessary which causes breast development and female appearance. The scrotal skin should be preserved during orchiectomy for urethroplasty. Consent of parents to change the child's gender should be obtained and psychosocial and legal problems solved (3,11). The management of aphallia that presents in later childhood and adulthood is a matter of greater controversy and difficulty. Here the type of treatment should be individualized (4). At present, our patient (Case 1) is five years old. His general condition and development is good and bears cystostomy without any problems. Renal and bowel function are excellent. Urethroplasty and vaginoplasty is planned for the future.

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