Penis, bladder and uretral agenesis associated with anorectal malformation in a living male neonate. Case report

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Summary

Aphallia is a very rare congenital malformation, with an occurrence of 1 in every 30 million births.

In the international literature about 75 cases have been indicated as of today.

The authors report and discuss the case of one neonate, born from a monoamniotic twin delivery, suffering from agenesis of the penis, anorectal malformation with a fully formed scrotum with 2 normal gonads and absence of bladder and urethra and both kidneys.

Introduction

Aphallia, albeit an exceedingly rare disorder with an occurrence estimated at 1 in 10 to 30 million births, must be managed in a judicious manner [1].

This includes exclusion of life-threatening anomalies, an accurate assessment of the urinary tract, early gender reassignment and immediate reconstruction.

The authors report a case of monoamniotic twins, one of them with agenesis of the penis, anorectal malformation, a fully formed scrotum with 2 normal gonads and absence of both kidneys, bladder and urethra.

Case Report

A neonate (weight of 2,160 gr.) born from a monoamniotic twin delivery, presented with penile agenesis associated with anorectal malformation, absence of the median rafe and normoconformed testicles and scrotum (Fig. 1 a-b). The child was the product of a 40-week gestation of a 36-year-old woman. The delivery was uncomplicated. There was no maternal history of toxin exposure or drug exposure with pregnancy. Family history was negative for congenital or familial disease. APGAR at birth was 10 on 10 and did not present any morphological alterations apart from those described.

The second twin (2,120 gr.) was perfectly normal with a slight form of hypospadia. Prenatal ultrasounds had shown a normal volume of amniotic liquid. Ultrasound at birth revealed absence of the kidneys and did not visualize the bladder. TC scan examination confirmed these findings.

Chromosomic investigation gave evidence of a normal karyotype 46 XY. The patient died of cardiocircular arrest at the age of 6 days without having had any kind of surgical operation. The autoptic exam confirmed not only the aphallia and the complete absence of the median rafe of the scrotum, but also the co-existence of bilateral renal agenesis and the presence of high anorectal malformation without fistula. Lung maturity was normal and there were no other associated malformations.

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Discussion

Agenesis of the penis is the result of complete or partial developmental failure of the genital tubercle [2].

At approximately 4 weeks' gestation mesenchymal cells develop about the cloacal membrane to form the genital tubercle as the urethral folds join in front of the cloacal membrane.

By 6 weeks the cloacal membrane is separated by the urorectal septum into a ventral urogenital sinus and a dorsal hindgut with anal membrane. During this period the urethral folds have divided into ventral urethral folds and dorsal anal folds. The genital swellings, lateral to the urethral folds, begin a caudal migration from the inguinal region to become the scrotum in the male subject.

The phallus, derived by elongation of the genital tubercle, draws the urethral folds forward with resultant formation of the urethral groove. By 12 weeks of embryogenesis the urethral folds unite over the urethral groove to form the portion of the penile urethra proximal to the glans penis.

Completion of the glandular urethra occurs during month 4 of gestation when canalization of the epithelial cord occurs to meet the pendulous urethra at the fossa navicularis [3].

The location of the urethral meatus at the anal verge represent failure of the urethra to develop beyond the most distal extent of the urogenital sinus. Although more distal urethral openings have been recognized they can be explained by partial formation of the urethral folds and groove [4].

More proximal urethral locations that communicate with the gastrointestinal tract most likely result from a combined abnormality of the genital tubercle and separation of the cloacal membrane by the urorectal septum. This combination occurs earlier in embryogenesis and, consequently, portends other associated anomalies.

Genitourinary anomalies have been seen in 54% of reported cases of aphallia and other system anomalies involving vascular anomalies and the gastrointestinal tract are reported [2, 5, 6].

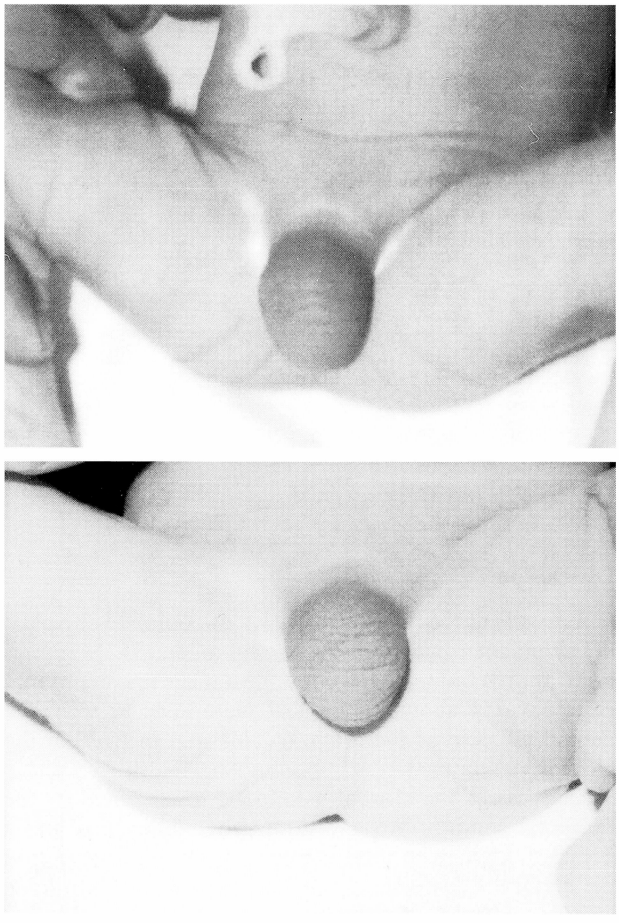

Figure 1. — **a)** The fully formed scrotum and testicles and the absence of the median rafe. **b)** The anorectal malformation and the absence of openings along the median line.

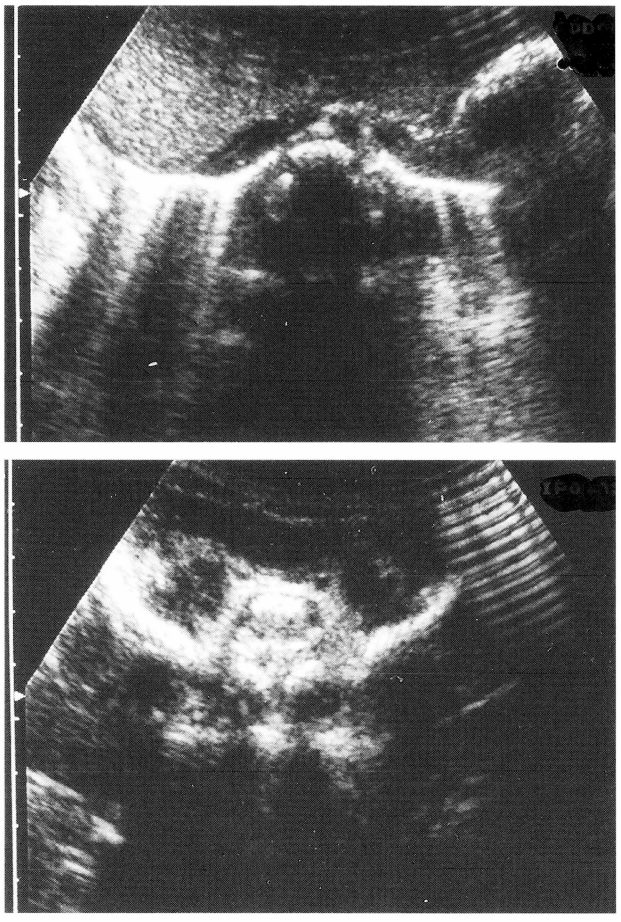

Figure 2. — Ultrasound shows the absence of both kidneys (**a**) and the absence of the bladder in front of the rectum (**b**).

In 1989 Skoog and Belman proposed a classification system for penile agenesis and noted that the specific position of the urethral opening was associated with prognosis [7].

We believe that the interest of our report is that an aphallic patient with bilateral renal agenesis had normal intrauterine life with no Potter Syndrome and a normal lung maturity at birth, due to the production of normal amniotic fluid by his twin's urine and mother's placenta.

In addition, to the best of our knowledge we believe that this is the first documented case of a live birth male neonate with the combined findings of penis, bladder and urethral agenesis associated with anorectal malformation.

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