

LETTER TO THE EDITOR

Anorectal Malformation with Absence of Penis, Bladder and Urethra

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DEAR SIR

Penile agenesis, or aphallia, is very rare and it is reported to present in one out of every 30 million live births [1]. Bladder agenesis is even rarer, with only 60 cases reported in the available literature, and only 19 of those cases were able to survive [2]. Here we are presenting a neonate with sequence of urorectal septum malformation with aphallia, bladder agenesis and anorectal malformation, expired on day two of life due to respiratory distress syndrome.

A 2.2-kg, first born, pre-term (32 weeks), neonate, from a consanguineous marriage referred from a primary health centre at day one of life. Though the pregnancy was unregistered but the mother had no antenatal complains. On examination of baby, phallus was absent; the scrotum was normal with two normally descended testes with palpable vas deferens. Urethral meatus could not be located anywhere in the scrotum or over the pubis. Anal opening was absent and gluteal cleft and anal dimple was not well developed (Fig.1). The neonate was in respiratory distress with tachypnea, tachycardia, central cyanosis, and bilateral coarse crepitation on auscultation. Abdomen was mild distended and soft on palpations.

Abdominal ultrasound was done that showed increased echogenicity in both kidneys, multiple simple cyst formations in both kidneys. In the pelvic cavity scan, no bladder, corpora cavernosa, or Müllerian structures were visible. The kidney functions of baby raised (serum urea- 60 mg % and creatinine-1.2 mg %). The patients planned for urinary diversion but due to respiratory distress syndrome intubated and kept on a ventilator and expired on next day.

The urorectal septum malformation sequence consists of absent perineal and anal openings in association with ambiguous genitalia and urogenital, colonic, and lumbosacral anomalies. The full sequence is highly lethal, and the partial sequence, characterized by a common cloaca, is compatible with life. Defects in mesodermal proliferation early in embryogenesis result in this rare condition. Aphelia is an anomaly which results from the non-formation of the genital tubercle during the fourth weeks of embryonic development accounting for its frequent association with related malformations. [3] In urorectal septum malformation sequence others associated malformations are common and include cryptorchidism, vesicoureteral reflux, horseshoe kidney, renal agenesis, imperforate anus, and musculoskeletal and cardiopulmonary abnormalities [4].



Figure 1 (A,B,C) showing absence of penis urethral opening and anal opening in a neonates.

Clinical presentation is diagnostic, but there are various other associated anorectal anomalies and other systemic anomalies which need to be investigated. Agenesis of the bladder is rarely compatible with life, only few living patients with this condition have been described.

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Our patient had the aphallia with absent bladder in addition to imperforate anus (complete urogenital agenesis was present), similar case also reported by Jose Manuel in that, high urinary diversion could not be done due to patient condition and expired postoperatively due to pneumonia [2]. In surviving patients, treatment goal is the sparing of renal function, with urinary diversion, antibiotic prophylaxis and electrolytic metabolic control. There used to be a general consensus that infants should undergo early gender assignment and be raised as girls, despite male karyotype [5].

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