Aphallia is a rare urogenital anomaly. It is a congenital absence of the penis. It occurs in about 1 in 500,000 births. It is associated with other developmental anomalies, including genitourinary abnormalities. The incidence of aphallia is estimated to be about 1 in 500,000 births.

Case Presentation

- A 33-year-old female
- Known as a boy
- Urinary drainage via a suprapubic tube
- Mild cleft palate
- Gastroesophageal reflux disease
- Asymptomatic intermittent right hydronephrosis
- No other significant anomalies
- Patient was born premature at 24 weeks gestation
- Diagnosis of aphallia was made at birth
- No other systemic anomalies on examination
- Bladder filling up first
- Proximal urethra with small diverticulum possible urocele
- Contrast passes into rectum

Introduction (Background)

Aphallia is a disorder of sexual development due to penile agenesis / congenital absence of penis. It is a very rare anomaly with only about 100 cases reported so far. Incidence is 1: 10,000 to 1: 30,000 births. Embryologically it is due to failure of the genital tubercle to develop during the 4th week of gestation. The penis is formed from epidermal pearls associated with other genital anomalies and most commonly a lower urinary tract fistula connecting to the rectum. Genitourinary anomalies are associated in up to 54% cases (including fistula, kidney and urethral abnormalities, VUR, absence of bladder and more). Aphallia is a sporadic anomaly usually associated with normal male karyotype and male hormonal metabolism. Absence of penis can be devastating to the parents and patient and may require multidisciplinary surgical and psychological management. Until the 1990's general consensus was to do a gender reassignment to female by early bilateral orchiectomy. Since the 1990's general consensus was to do a gender reassignment to female by early bilateral orchiectomy to decrease masculinization effect by testosterone. To prevent urinary infections the urinary and local problems were separated at early age. And vaginoplasty and estrogen therapy to be done in adult life. New guidelines as per Intersex Society of North America: If normal testes, declared male sex. If normal ovaries, declared female sex of rearing and Y chromosome, 53% later with disorders of sexual development. Of 60 patients with the declared sexual identity to sex of rearing in individuals with disorders of sexual development. Of 60 patients with the declared sexual identity to sex of rearing in individuals with disorders of sexual development. Of 60 patients with the declared sexual identity to sex of rearing in individuals with disorders of sexual development. Of 60 patients with the declared sexual identity to sex of rearing in individuals with disorders of sexual development.

Management is controversial. A study by Reiner et al. compared the development of children with aphallia in individuals with disorders of sexual development. Of 60 patients with female sex of rearing and 9 chromosome, 53% later declared male sexual identity. Of 2 aphallia patients reared as female, 1 declared male identity later. 15 patients reared as males, declared male identity. Spontaneous declaration of male sexual identity was an early age of 4.5 years. DeCastro et al. propose early temporary phalloplasty using an abdominal wall skin flap in childhood followed by definitive phalloplasty with labial skin flap from an area. Many surgical approaches to phallicplasty have been described around the world. But it should be recognized that penis reconstruction is demanding and often requires multiple revisions.

Results (with Management)

- Imaging:
  - KUB: Generalized distension of bowel with gas noted in rectum.
  - No vertebral anomalies.
  - Ultrasound: Kidneys are grossly normally bilateral.
  - Bladder can be visualized in the same suprapubic location but there appears to be a collection of fluid just inferior to the umbilicus.

Abstract

Aphallia is a rare urogenital anomaly with less than 100 cases reported in literature and co-exists with other genitourinary anomalies that require immediate attention. It also has dramatic psychological consequences. Diagnosis can be established by physical examination, karyotyping and radiological investigations like Xrays, VCUG and MRI if needed to guide anatomical interpretation. Management includes, separation of the urorectal fistula along with a phalloplasty.

Case Presentation

- 33 year female, GUQ50 congenes by in vitro fertilization. Also has hypertension being treated with Metoprolol. At 19.6 weeks AFP screen is positive. A follow-up at 20.6 weeks USG was done which revealed singleton male fetus, distal bowel cystic present at insertion of umbilical cord or within cord, 2 vessel cord and markedly decreased Amniotic fluid index (5 percentile). Mother refused Chronic Villus Sampling. For Chromosomal analysis. Subsequent USG’s however only revealed persistence of 2 vessel cord and bilateral renal pyelectasis. Generalized distension of bowel with gas noted in rectum.

References