ABSTRACT

Aphallia or penile agenesis (PA) is an extremely rare genitourinary anomaly and an rare form of genital ambiguity with an estimated incidence of 1 in 10–30 million livebirths. The earliest case report of aphallia was by Imminger in 1853. Penile agenesis (PA) is believed to result from either the absence of the genital tubercle or its failure to develop. Patients usually have 46XY karyotype. More than half of these have associated anomalies, including developmental defects of the caudal axis, genitourinary and gastrointestinal tract anomalies. The scrotum, testes and testicular function are usually normal. Patients should be genetically evaluated at birth and associated malformations should be looked for. It is important to propose early sex reassignment. Surgical intervention during infancy, including urethral transposition and bowel vaginoplasty via a posterior sagittal approach plus orchiectomy and scrotoplasty, has been advocated. However, gender reassignment in other contexts and detailed psychosexual data may not support this recommendation. We report a case of a aphallia in a neonate born at a tertiary hospital MIMS Mandya.

KEYWORDS
Aphallia; Penile Agenesis, Genital, Ambiguity.

INTRODUCTION

Aphallia or penile agenesis is an extremely rare genitourinary anomaly with an estimated incidence of 1 in 10–30 million.[1] After the first case report was described in 1853 by Imminger, less than 100 cases have been reported worldwide in the English literature. PA is believed to result from either the absence of the genital tubercle, or its failure to develop.[2] Several investigators claim the absence of corpora cavernosa and corpora spongiosum as a prerequisite for the diagnosis of penile agenesis. patients have 46 XY karyotypes. More than half of these have associated anomalies, including developmental defects of the caudal axis, genitourinary and gastrointestinal tract anomalies.[3] In the majority of cases the urethral meatus is somewhere in the perineal region or forms a fistula to the gastrointestinal tract, usually towards the rectum. There is greater associated mortality and malformations when the meatus is more proximal.

The aim of treatment is an early female gender assignment and feminizing reconstruction of the perineum.[3] Herein, we report a case of an aphallia in a neonate born at a tertiary hospital MIMS Mandya.

CASE REPORT

A 21-year-old female with G1P1L0 34 wks of gestation with history of consanguineous marriage admitted to admitted to labor ward with labour pain, underwent LSCS delivered a baby with birth weight 1.6 kg with aphallia (Fig-1) and urination and defecation through rectum. Physical examination revealed absence of penis, apparently normal scrotum, and bilateral well descended testes. Anus was located on its normal place (Fig-2) with no urethral opening. No history of exposure to toxic substances or X-ray, or medicine intake during his mother’s pregnancy was obtained.

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Ultrasonography abdomen and pelvis reported normal study, urinary bladder present. USG cranium was normal, 2D ECHO and chest x-ray were normal. Karyotyping, 17OHP reports awaited. Attendants were explained about further treatment and planned for perineal urethrostomy after 6 months.
DISCUSSION
Aphallia or penile agenesis is a severe genitourinary tract malformation and is believed to be caused by the complete or partial maldevelopment of the genital tubercle. It should be differentiated from some congenital deformities, such as severe hypospadias, severe epispadias, intrarterine penile amputation,pseudohermaphroditism, concealed penis and micropenis. A total of 54% of cases of penile agenesis are found to be associated with genitourinary anomalies. This anomaly was first described by Imminger in 1853. Since then approximately 100 cases have been reported worldwide.[4,5]

During week 3 of embryogenesis, mesodermal tissue from the region of the primitive streak migrates down about the cloacal membrane to form the cloacal folds. These folds unite in front of the cloacal membrane and form genital tubercle. While this occurs externally, the cloaca is divided internally into the anterior urogenital sinus and posterior rectum by the urorectal septum. As the embryo matures, the genital tubercle elongates and the cloacal folds divide into the anterior urethral and posterior anal folds. The genital swelling appears at this time on either side of the cloacal folds, these swellings eventually will migrate caudally, coming together over the urethral folds and fusing at the median raphe to form the scrotum. The genital tubercle will fuse to form the urethra internally and the penis with its median raphe externally.[6,7] Penile agenesis is believed to be due to deficient formation of the genital tubercle or its failure to develop into a penis in the fourth week of embryogenesis.

Skoog and Belman[3] suggested three variants, based on urethral position in relationship to the anal sphincter, as: Postsphincteric, Presphincteric (Prostatorectal fistula) and Urethral atresia. More proximal the bladder outlet, greater is the likelihood of other anomalies and death.

The diagnosis of this abnormality, includes: complete absence of corpora cavernosa and corpus spongiosum and opening of the urethra in perineum near the anus or into the rectum.[8,9]

Treatment for aphasis should be initiated as early as possible, based on careful evaluation and prompt gender assignment by a clinical team, including a psychologist, an endocrinologist and a urologist. For infants with aphasis, the recommended surgical treatment for patients with aphasis is early sex assignment to the female gender. Many paediatricians advocated bilateral orchiectomy and neovaginal reconstruction during the neonatal period, and oestrogen therapy for breast development and other female sexual characteristics[10] However, for patients who delayed early intervention, total phallic reconstruction might be another alternative considering the preference of patients as well as the prevailing socioeconomic conditions.

CONCLUSION
Congenital aphasis is an extremely rare condition that requires multidisciplinary and individualized management. The most accepted management is early sex reassignment with correction of obstructive uropathy. In newborn period or infancy, feminizing operations are recommended for treatment of PA, but after two years, as sexual identification of the patients has appeared, it is advised to perform masculinizing operations in order not to disturb the patients psychologically, and finally, no surgical intervention should be performed before counseling the parents.

REFERENCES