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Research Article

Aphallia: Scenario in Bangladesh and Its Management

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Abstract: Aphallia, congenital absent of penis, is a rare, urogenital malformation of the external genitalia in a genetically male child (karotypically XY). Even though the exact cause is unknown but it is may be due to an awry in development of external genitalia a child. Incidence of aphallia is 1 in 30 million live births. We are presenting 03 cases of aphallia associated with other congenital malformations. This rare problem needs thorough evaluation for possible associated malformations and need multistage and multidisciplinary management.

Keywords: Aphallia, congenital malformations.

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INTRODUCTION

Aphallia or penile agenesis is an extremely rare genitourinary anomaly with an estimated incidence of 1 in 10–30 million. [1] Aphallia was first reported by French Surgeon, Saviard in 1701, however detailed description was given in 1853 by Imminger. To the date, around 100 cases have been reported worldwide [2]. Aphallia is believed to result from either the absence of the genital tubercle, or awry in the development of formation of penis. [3] Several investigators claim the absence of corpora cavernosa and corpora spongiosum as a prerequisite for the diagnosis of penile agenesis in a genetically male child (46 XY karyotypes). More than half of these have associated anomalies, including developmental defects of the caudal axis, genitourinary and gastrointestinal tract anomalies. [4] 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.

CASE 1

A 6-month boy presented with acute retention of urine in emergency department with absence of phallus which relief by cutaneous vesicostomy. After birth, the baby passes urine through a ectopic urethral opening present in anterior wall of anus with defecation but without difficulty. On examination there is no phallus but pit of tissue present, well developed scrotum with testes and no other associated anomalies. Karyotyping revels 46 XY. Ultrasonography of whole abdomen shows normal. X-ray of lumbosacral spine normal. Urethral repositioning was done in perineum outside the anal sphincter at the age of 4 years. From which he now void normally. Episodic UTI occur.

Figure 1: case 1

CASE 2

A 3 months' boy attending emergency department with acute urinary retention and acute abdomen. After resuscitation, exploratory laparotomy followed by colostomy and cutaneous vesicostomy was done. After 2 years, again refashioning of vesicostomy was done. At birth absence of penis with well-developed scrotum and palpable testes. Urethral opening was present in rectum. The baby passes gushes of urine during defecation with continuous wetting of napkin. Karyotype

shows 46 XY, ultra-sonogram of whole abdomen and x-ray lumbosacral spine revels normal. Child is now passes stool through colostomy and urine passes through anal opening.

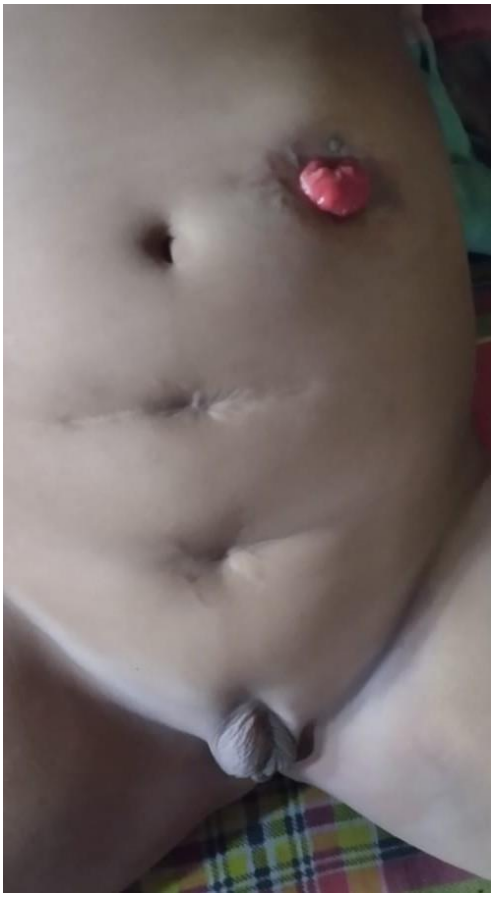

Figure 2: case 2

CASE 3

A 6 years' boy presented to me with recurrent UTI having aphallia. Parents were worried regarding their child future. On examination urethral opening in the perineum just above the anterior wall of anal opening which was erythematous. Urine RME shows significant pus cells. Ultra sonogram of abdomen shows right ectopic kidney with no radiological spinal abnormalities. Karyotyping is 46XY.

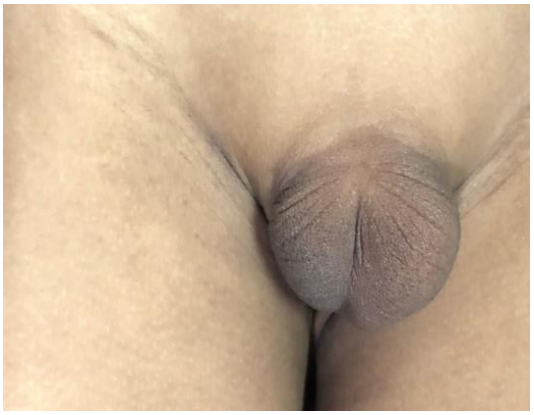

Figure 3: case 3

DISCUSSION

The causes of penile agenesis are unknown. It is thought that, it occurs as a result of absence of genital tubercle or deficiency in its development with incomplete separation of the urogenital sinus from the hindgut by the urorectal septum. [2,5] Diagnosis of aphallia includes the absence of phallus, male karyotype and normally developed scrotum and normal and frequently undescended testes. [2]

Aphallia may occur alone or it may have associated other congenital anomalies. Penile agenesis associated with other malformations is much more prevalent (54%) than the solitary variety. Aphallia is a part of urorectal septum malformation sequence (URSMS) and are associated with genitourinary, cardiac, gastrointestinal, nervous system and musculoskeletal system anomalies. Genitourinary anomalies include renal agenesis, renal hypoplasia, renal dysplasia, hydronephrosis, ureteral agenesis, ureterocele, single testes, Cryptorchidism. Cardiac anomalies include co-arctation of aorta, tetralogy of fallot, truncusarteriosus with ASD, VSD and right sided descending aorta. Among gastrointestinal anomalies are tracheoesophageal fistula, malrotation of gut, anorectal malformation and rectovaginal fistula. Nervous system anomalies include lumbar spine dysgenesis, sacral dysgenesis, myelomeningocele and arachnoid cyst. Penile agenesis may also have associated musculoskeletal anomalies such as hypoplastic leg, flexion contractures of hip and knees, tibial bowing and bilateral equino-varus of feet. [6] Other anomalies such as Cleft lip, Cleft palate and Prostate agenesis may also be present with aphallia. [7] Here 3 cases are different types and presentation. In Case 1, ectopic urethral opening present in anterior wall of anus and presentation was retention of urine. In case 2, ectopic urethral opening present in rectum and presented with acute abdomen and acute urine retention in last case, ectopic urethral opening present in perineum. All cases presented with feature of UTI. one case presented with ectopic kidney with no other associated anomalis, all parents were worried about their sexual life. So, a comprehensive evaluation of all systems is required in patients who present with aphallia to rule out all such anomalies. Management of aphallia is controversial. Apart from the aforementioned associated congenital anomlaies, aphallia should be differentiated from other similar conditions like micropenis, rudimentary penis, penile amputation, concealed penis and pseudo hermaphroditism. [5] Genital reassignment surgery is the main stay of treatment. There is a need for complete assessment of patient's clinical, psychological and social factors before gender assignment. Some do feminizing genitoplasty (Bilateral orchiectomy with vaginal reconstruction, labial reconstruction and urethral transposition followed by life-long hormonal therapy initiated during puberty) [8] while others believe that penile reconstruction (phalloplasty) [3] is a good option. But in some religious perspective people are not ready for gender reassignment Surgery. So, there is great dilemma between both the treatment modalities. The newer concept of gender imprinting in brain since in-utero

and the psychosocial consequences of gender reassignment is an important perspective in such cases. [9] Thus the treatment should be individualized according to age of presentation, psychosocial adjustment of both patient and parents as well as socio-religious context. The treatment is a multidisciplinary approach involving a expertpaediatric surgeon, urologist, endocrinologist and psychologist.

CONCLUSION

Aphallia is a rare condition that may have associated congenital anomalies. A comprehensive approach is necessary for evaluation and management of these cases. Thus, treatment should be individualized depending on the age of presentation and psychosexual orientation of patient and also socio- religious context. Pre-surgical counseling remains an indispensable part of the treatment.

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