CASE SERIES

APHALLIA: A VERY RARE CONGENITAL ANOMALY, WITH ASSOCIATED GENITOURINARY AND ANO-RECTAL MALFORMATION

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Aphallia or penile agenesis is very rare congenital anomaly of unknown cause occurring 1 in 30 million live births. Very little has been written in literature about aphallia. There is absent phallus and urethra may open abnormally in perineum or into rectum posing various surgical, social and psychological implications as the child grows. We are presenting 03 cases of aphallia with associated congenital anomalies such as unilateral renal agenesis, bilateral undescended testes, anorectal malformation and rectovasical fistula.

Keywords: Aphallia; Penile agenesis; Absent penis; Ambiguous genetalia; Phalloplasty; Neo-phallus; Penile reconstruction.

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INTRODUCTION

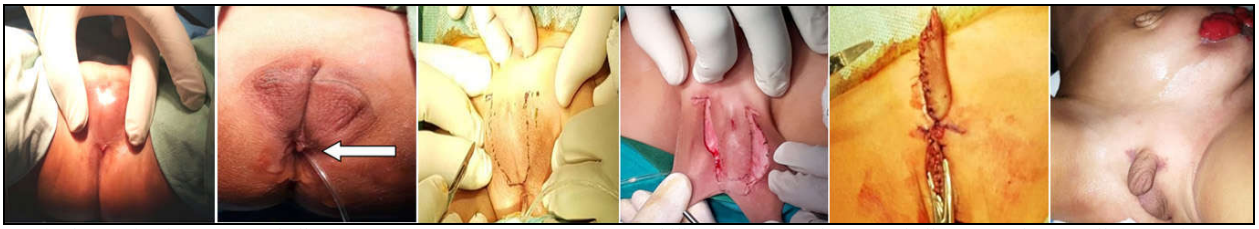
Aphallia or penile agenesis is a rare congenital malformation in which penis or clitoris is absent. It is very rare anomaly occurring once in 30 million live births. It was first described by Imminger in 1853. Nearly 100 cases have been reported worldwide so far.[1]

CASE-1

Two and a half years old baby born through normal vaginal delivery born full term was referred to our hospital on 2nd day of life with complaints inability to pass meconium. Examination of the child revealed no anal opening, absence of phallus, urethral opening was present in perineum posterior to scrotum and there was under developed scrotum with both testes not palpable in scrotum. Rest of general and systemic physical examination was normal.

Pelvic divided colostomy was made for anorectal malformation and child remained in hospital for seven days and discharged with advice of follow up for further workup of aphallia. Karyotyping showed 46XY with no gross chromosomal anomaly.

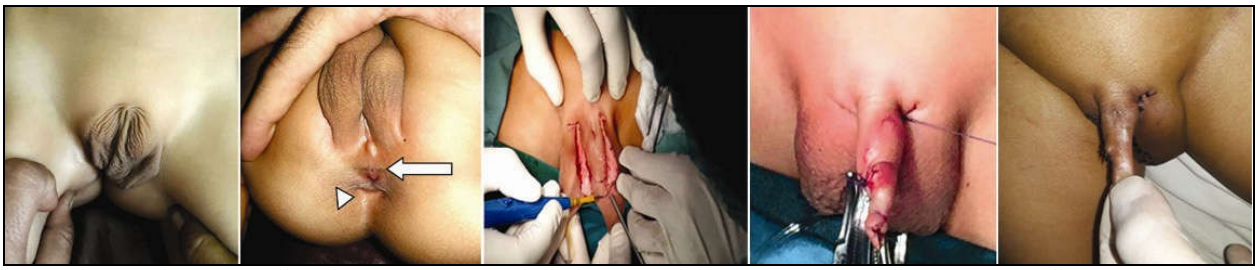
Ultrasound revealed absence of left kidney in abdomen and both testes present bilaterally in both inguinal canals. MAC-3 renal scan showed 100% functioning of right kidney with non-visualization of left kidney throughout the scan. Cystoscopy revealed normal bladder. Echocardiography was done and there was no structural and functional anomaly with ejection fraction of 80%. Phalloplasty was done using scrotal skin flap. Our plan is to do staged orchidopexy, and transposition of urethra to neophallus at later stage.



Single opening in perineum | No phallus (arrow) Soft tube in urethra | Incision on scrotal skin for phalloplasty | Mobilization of scrotal skin | Neophallus (ventral view) | Follow up after 8 weeks

CASE-2

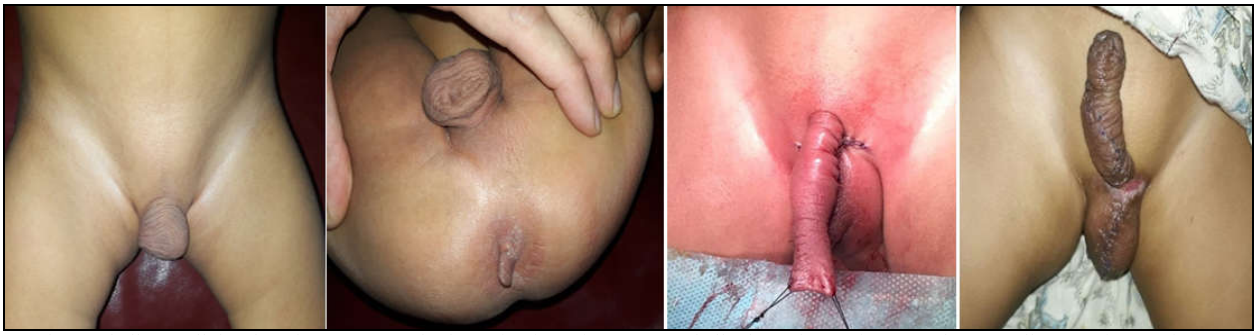
Four years old child born full term through normal vaginal delivery presented in the OPD as a case of ambiguous genitalia with absent penis and child is passing urine through an opening in perineum. On examination, there was absent penis. Child has well developed scrotum with bilateral testes palpable in scrotum. There was normal anal opening. Urethra was opening in the perineum between scrotum and anus. Other systemic examination was unremarkable. Ultrasound abdomen and pelvis was done which showed no sonographic evidence of uterus or ovaries. Further workup showed 46XY karyotype. Renal scan showed fair functioning of bilateral kidneys with dilated non-obstructive right pelvicalyceal system. Scrotal phalloplasty was performed in this child.



Absent penis | Bilateral testes present, urethral opening (arrow) anus (arrowhead) | Phalloplasty, scrotal skin incision with mobilization of flap | Neophallus | Follow up at 04 weeks

CASE-3

10 months old child presented to us with complaints of absent penis. Child was passing urine through anal opening. On examination, there was absence of phallus, bilateral testes were palpable in scrotum. No urethral opening was found in perineum. On investigation child had 46XY karyotype. Ultrasound abdomen revealed non-visualization of right kidney. Renal scan was performed which showed absent/non-functioning right kidney and fair functioning, hydronephrotic left kidney with satisfactory excretion and there was left hydroureter. Renal function tests were normal. Intravenous urogram was done which showed absent right kidney, left hydronephrosis and hydroureter with rectovasical fistula. Phalloplasty was performed using scrotal skin flap.



Absent phallus, well developed scrotum | No urethral opening in perineum, anal opening present | Phalloplasty done using scrotal skin flap | Follow up picture after 4 weeks

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.[1,2] Diagnosis of aphallia includes the absence of phallus, male karyotype and normally developed scrotum and normal and frequently undescended testes.[1] our first case report has aphallia with bilateral undescended testes. The urethra opens anywhere in midline in the perineum between pubis to anus and sometimes in anterior wall of rectum. Our second case has urethral opening in perineum between scrotum and anus. Based on position of urethra to anal sphinchter, Skoog and Belman classified aphallia into three categories: presphenchteric, post sphenchteric and urethral atresia.[2]

Aphallia may occur alone or it may has associated other congenital anomalies. Various associated congenital anomalies have been reported in literature. Sharma et al described aphallia as a part of urorectal septum malformation sequence (URSMS) and associated anomalies include genitourinary, Cardiac, Gastrointestinal, Nervous system and Musculoskeletal system anomalies. Genitourinary anomalies include Renal agenesis, Renal hypoplasia, Renal dysplasia, Hydronephrosis, Ureteral agenesis, ureterocele, Mono orchid, Cryptorchidism. Cardiac anomalies include AV canal defects, Coarctation of aorta, tetralogy of fallot, truncus arteriosus 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 has associated musculoskeletal anomalies such as hypoplastic leg, flexion contractures of hip and knees, tibial bowing and bilateral equinovarus of feet.

Other anomalies such as Cleft lip, Cleft palate and Prostate agenesis may also be present with aphallia. So, a comprehensive evaluation of all systems is required in patients who present with aphallia to rule out all such anomalies.[3,5]

Other differentials of aphallia include concealed penis, rudimentary penis, micro penis, intra uterine amputation of penis, epispidias, hypospedias and pseudo hermaphroditism.[2]

Management of penile agenesis (Aphalia) is controversial. There is a need for complete assessment of patient's clinical, psychological and social factors before gender assignment. Some do feminizing genitoplasty while others believe that penile reconstruction (phalloplasty) is a good option. Ideally female gender should not be assigned to patients with aphallia and they should be raised according to their male karyotype and hormonal production because majority of patients show male typical shift in their psychological, social and sexual behavior after long term follow up.[1,4] Male gender assignment consists of reconstruction of the penis that is phalloplasty. In female gender assignment bilateral orichidectomy, urethral perineal transposition and feminizing genitoplasty is done before testosterone surge. Vaginoplasty is done at latter stage. Female gender assignment is easier but requires lifelong hormonal therapy with estrogens so that they can have secondary sexual characteristics. Hormonal therapy is started at puberty.[5] In our part of the world penile reconstruction is a better option due to psychosocial Implications.

CONCLUSION

Aphallia is a rare condition that may have associated congenital anomalies. A comprehensive approach is necessary for evaluation and management of these cases. Phalloplasty is the ideal option to overcome psychosocial problems.

AUTHORS' CONTRIBUTION

MUN: Conceptualization of article design, write-up, literature search. NA: Article design, article revision and proof reading, final approval of the article. MFG: Editing and literature search. SS: Editing and Literature search. SV: Literature search

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