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Surgery

CAUDAL DUPLICATION SYNDROME: CASE SERIES AND REVIEW OF LITERATURE.

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ABSTRACT Caudal duplication syndrome is a rare entity in which structures derived from the embryonic cloaca and notochord are duplicated to various extents. There have been isolated reports of duplication of the colorectum, lower urogenital tract, spinal dysraphism and abdominal wall defects resulting from insults at different stages of embryogenesis. We herein describe 3 cases of diphallia, one case of anal duplication, one case of hindgut duplication and one extremely rare anomaly of labial duplication showing corporal tissue which has never been previously reported. The management of these rare anomalies is individualized and literature reviewed. By combining these rare cases together with similar embryological background we discuss the pathological anatomy, management, results of our experience.

KEYWORDS : .Diphallia, Anal duplication, Vulval duplication, Hindgut duplication.

Introduction

Caudal duplication results from sagittal symmetric pairing of axial structures of the caudal embryo. In its complete form, it comprises of duplicated colorectum, and anal canal with double anal orifices; two hemibladder, urethra, phallus; infraumbilical abdominal wall defects including omphalocele, vesical extrophy, and pubic diastasis, and additionally in girls, 2 unicornuate uterus, double vaginae, and vulvae. Presence of double vertebral column is also a feature in this syndrome. The embryological basis for the malformations seen can be explained by an insult to the caudal cell mass at day 23-25 of gestation resulting in caudal duplication syndrome, it is also referred to as split notochord syndrome or dipygus.[1] In our institute we have managed 6 cases of partial caudal duplication and have achieved good functional results.

Material and Methods

A total of 6 children with a varying degrees of caudal duplication were treated at our institute, from 2010 to 2012. Individualized investigations for every case were done.

There were 3 males and 3 females. Age ranged from 3months to 3 years. All patients were evaluated accordingly by ultrasonography (USG), micturating cystourethrogrm (MCU), sonogram and cystoscopy.

The clinical presentation and associated anomalies are summarized in table 1.

Sr. No	Case	Age/ Sex	Clinical Presentation	Associated features	Treatment	Follow up and result
1	Incomplete Diphallus	2years male	2 streams of urine and absent anal opening	Anteposed stenotic anus	Diphallus correction and PSARP	2 year and passing urine with good stream
2	Diphallus	1 year male	Absent anal opening and double phallus, three hemiscrotum and lipomatous mass	Anorectal malformation, Meckels diverticulum, malrotation, perineal lipoma, complete urethral duplication	Meckels diverticulectomy, correction of the malrotation and divided transverse colostomy f/b final surgery	2 year follow up is asymptomatic
3	Incomplete Diphallus	3 years male	Retention of urine suspected to have urethral atresia	Double bladder, single functioning kidney with VUR, complete urethral duplication, CRF	Right ureterostomy	Awaiting final surgery
4	Anal Duplication	3 years female	Discharge from sinus	None	Excision of accessory tract	Asymptomatic
5	Labial duplication	3mths Female	Absent anal opening at normal site with vestibular fistula	Low ARM	ASARP and excision of accessory labia	Continent, asymptomatic
6	Hindgut duplication	4mths female	Passing feces through two opening in perineum		Split sigmoid colostomy	Awaiting definitive surgery

Case 1-3 were cases of diphallia. Case 1 was case of glandular diphalllia (fig. 1-2) with associated anteposed stenotic anus. Micturating cystourethrography revealed well-formed ventral urethra and atretic dorsal urethra with normal bladder and posterior urethra. MRI was done to delineate the anatomy and revealed a well formed corpora cavernosa of dorsal penis and only corpora spongiosa in ventral duplicated glans. Genital reconstruction was done with meatotomy of the ventral urethra and ventral urethra with spongiosa was placed in between the corpora cavernosa of dorsal phallus.

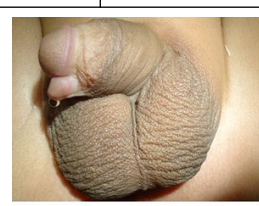

Fig 1: Duplicate glans with a drop of urine

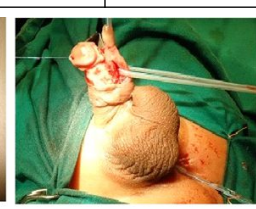

Fig 2: Ventral atretic glans and spongiosa

Figures 1-2: Glandular diphallus.

Case 2 had double penis, three hemiscrotum and lipomatous mass at base of scrotum. Patient was passing urine in two stream. Patient had associated high anorectal malformation which was managed in staged manner. For genital reconstruction, MCU was done by catheterizing both urethra which revealed double anterior urethra and single bladder. Accessory penis and scrotum along with the lipomatous mass was excised. Postoperative patient is asymptomatic.

Case 3 was complex case of glandular diphallia with ventral atretic urethra (fig 3) and single functioning right kidney with right grade 3 VUR. Child has underwent right ureterostomy and is awaiting follow up surgery.

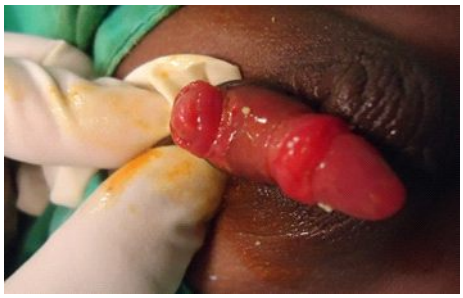

Figure 3: Glandular diphallia with ventral atretic urethra

Case 4 was 3 year old female had small opening measuring 4-5mm in diameter located just posterior to the anus in the midline had been noted since birth but ignored. Anus was normal in position with normal sphincter tone and anal function. No bony defects were present. There was no history suggesting infection of the sinus. USG pelvis was normal. A complete excision of blind ending tract through anterior approach was done. Histopathology showed squamous epithelium with smooth muscle and anal ducts around the cavity.

Case 5 was a 5 months old girl presented with history of passage of stool through abnormal site. On physical examination, the most surprising sign was accessory mass on the left side of labia majora. Three openings were present in the perineum including urethra, vagina and vestibular fistula. There were no associated urinary, cardiac or bony abnormalities. Complete excision of accessory mass an ASARP was done. Histopathology showed presence of labial tissue with clitoral tissue thus suggesting it to be a labial duplication.

Case 6 was 4 month old baby presented with fecal matter coming through 2 openings in the perineum. Examination under anaesthesia revealed normal anus and a vestibular opening with no communication between the two (fig 4). A laparotomy a diversion was planned in view of complex anomaly. At laparotomy for sigmoid colostomy, a Y-shaped tubular duplication of the transverse, descending, and sigmoid colon was encountered with both limbs leading to the pelvis (fig 5 and 6). The remaining colon and abdominal viscera were normal. A split sigmoid colostomy was fashioned in the left iliac fossa, resulting in 4 stoma openings. Postoperatively, both proximal lumens drained feces. The child is awaiting follow up and final corrective surgery.

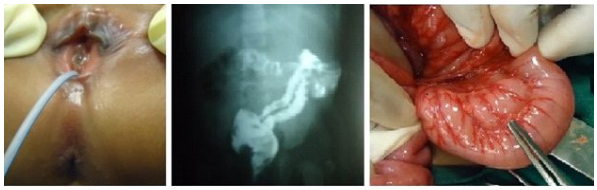

Fig 4: Rectovestibular fistula with normal anus

Fig 5: Contrast study showing duplication of colon

Fig 6: Duplicated sigmoid colon

Figure 4-6: Sigmoid colon duplication.

Discussion

The caudal end of the human embryo gives rise to the primitive urogenital sinus, anorectum, and cloaca. For nearly 100 years it was taught that this caudal end of the embryo was partitioned by the fusion of the lateral walls of the cloaca.[2] and cranial-caudal descent of a septum: the urorectal septum.[3] Together these embryological events bisected the cloaca into an anterior, primitive urogenital sinus, and a posterior anorectum, between the fourth and sixth weeks of embryogenesis. Moreover, it has been assumed that the distal end of the sewer or cloaca somehow fused with these separate tubes and formed openings for these systems to the exterior. These seminal theories have been modified to include a single septation[4, 5] or consecutive septations.[6,7] It is now accepted that, in humans, a separate primitive urogenital sinus and an anorectum exists as early as the fourth week of gestation. Moreover, the cloaca serves as the channel to the exterior for each of these orifices when the cloacal membrane dissolves. There appears to be no evidence for a descent of the urorectal septum and, thus, septation of the cloaca. The first detailed and systematic illustrations of human embryos were done by Wilhelm His at the University of Leipzig in 1885.[8] The Carnegie Embryological Collection, which contains serial histological sections of human embryos and fetuses is the standard reference for staged human embryos.[9]

Diphallia is rare genital duplication with incidence of 1 in 5 million births.[10] Associated congenital anomalies are present in the majority of cases. In our study we had imperforate anus (case 1 and 2), colon duplication (case 2), duplicated bladder and urethra (case 3).This is different in reported cases.[11,12,13] Intestinal anomalies are mostly associated with complete diphallia, and imperforate anus.[14,15] We had only one case of this combination in our series. Treatment of diphallus usually includes excision of the duplicated penile structure and its urethra. Associated anomalies can also be repaired surgically.[15,16,17]

Anal canal duplication (ACD) are rare with only 50 cases reported in world literature. Maximum of reported cases are in girls as also in our case.[18] The length varies from 10-30 mm, in our case it was 15mm. ACD are usually asymptomatic but early diagnosis can be made by simple perineal examination. Associated anomalies reported in literature are dermoid cyst, presacral teratoma, ureteral duplication, lumbosacral meningomyelocele and spina bifida, [19] none were present in our patient. Hamada et al proposed the pathogenesis of ACD may be explained by duplication of dorsal cloaca in early developmental stage of 30 day old embryo.[20] Histology of ACD may show a variety of different epithelia. Most commonly squamous or transitional epithelium with smooth muscle components and anal ducts is present and was seen in our case too. Treatment is complete excision which is required to avoid complications like accumulation of secretions and infection.

Double vulva is also an extremely rare congenital malformation, and there are only about 20 previously reported cases in the world literature.[21] Although the genital system is closely related with the urinary system and shared with many common sources in their embryological development, the internal genital organs (ovary, uterus, and vagina) have different embryological sources from the vulva (vestibule, clitoris, and labia minora and majora).However isolated labia majora duplication with clitoral duplication with anorectal malformation has not been reported in world literature. Simple excision is only treatment required and we have achieved good cosmetic and functional outcome in this rare case.

Duplications of the gastrointestinal tract are rare congenital malformations with an incidence of 1:4000 -5000.[22] Colonic duplications represent 3% to 20% of all gastrointestinal duplications.[23,24] Tubular hindgut duplications are associated with genitourinary anomalies in 60% of cases.[25] Fistulae between the duplicated colon and the genitourinary system or perineum are present in 50%.[25] The presentation of anorectal malformation and associated hindgut duplication is both uncommon and confusingly classified in the literature. A practical classification based on the status of both the colon proper and the duplicated colon can be useful. Each colon may be associated with a normal anus, a fistula, or it may be blind ending. This results in 9 possible permutations, accepting the obvious variability of the fistula(e). There are two options for management of the more extensive proximal duplicated segment. The first, and the option described in this report, involves resection of both limbs to leaving a single proximal transverse colostomy. Resection has major implications for final colonic length and potential future continence, particularly where the origin of the duplication is very proximal, but it does eliminate any concerns related to ectopic mucosa. Alternative options include performing mucosectomy on one limb of the proximal duplication or retaining both limbs with a distal stapled approximation to provide a single defunctioning colostomy to protect the anorectal reconstruction. In presence of common mesentery, the proximal duplicated limb should probably be preserved, especially if extensive, to optimize the functional outcome of the anorectal reconstruction.[26]

CONCLUSION

Partial caudal duplication can have varied presentation. By discussing management of 6 rare cases we conclude that proper search for the associated malformations in such rare cases is important in complete surgical management and in achieving optimum results. By clubbing such rare cases with common embryological association we would like to suggest that thorough evaluation is the key to successful outcome in such cases.

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