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Cloacal Exstrophy: A Case Report and Literature Review

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Summary. *Cloacal exstrophy is an extremely rare congenital malformation resulting in an exstrophy of the urinary, intestinal, and genital organs and associated with anomalies of other organ systems. We present a complicated case of cloacal exstrophy and the recent progress in the management of this probably most complicated anomaly in pediatric urology and surgery.*

Introduction

Cloacal exstrophy, sometimes also called vesicointestinal fissure, is probably the most challenging birth defect in urology and pediatric surgery. It was suggested even to relate the number of departments of pediatric urology in a country to the number of newborns with exstrophy born per year. Patient survival and optimal management of this anomaly reflect the maturity of pediatric surgery and urology and pediatric health care in general. Since every case is different in a complexity of the anomaly, we present our case with review of the literature.

Cloacal exstrophy is an extremely rare congenital disorder thought to be related to abnormal development of the cloacal membrane, a transitory structure composed of endoderm and ectoderm that overlies the embryonic cloaca. The arrested development of the cloaca leads to urethral, vaginal, and rectal openings all sharing a common single external orifice. The faulty embryogenesis in cloacal exstrophy compounds the problem by rupture onto the anterior abdominal wall. The result is primarily an abdominal wall defect with failed closure of the lower urinary tract, maldevelopment, and failed closure of the colon. Several theories have been suggested, but the exact nature of this problem is currently unknown.

Over the last 30 years, the focus has shifted from the survival, despite its complexity, to improving patient outcomes and ensuring the optimum quality of life for him/her afterward, which includes an appropriate gender assignment, independence from stoma appliances, higher level of physical and social independence and mobility. The priority is given to urinary, gastrointestinal, and genital reconstruction, designed to adapt the patient as a free person and of appropriate psychological gender.

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Case Report

A 24-year-old woman gave birth to a preterm alive newborn (gestational age, 32 weeks). This was the second pregnancy and the first delivery. The first pregnancy was terminated due to complications at the eighth week of pregnancy. She did not have any antenatal history of drug intake or infection. Antenatal ultrasonography revealed the fetus in the breech position having an omphalocele with no visualization of the bladder. The newborn was born by cesarean section. Birth weight and height of the fetus was 1930 g and 41 cm, respectively. The mother, together with her child, was transferred to a tertiary center, i.e., a neonatal intensive care unit.

On external examination, the ventral abdominal wall was found to be defective with a giant omphalocele incorporating the liver, stomach, spleen and loops of intestine, covered by a partially ruptured membrane with exstrophy elements in the lower part (Figs. 1 and 2). Male sex was confirmed after examination of the external genitalia. Normal 46 XY karyotype was determined later. Just above the epispadiac glans penis, there was a duplicated bladder patch with 2 ureteric orifices separated by an extrofied cecal patch with 4 orifices. On squeezing the abdomen, a speck of meconium was found to appear in the upper part of the central mucosal patch. There was an imperforate anus with only a dimple seen in the perineum. The penis was epispadiac. On x-ray, sacral agenesis and spina bifida was detected. Further examination of the pelvic organs with sonography showed the pelvic hypoplastic left kidney and intraabdominal testicles.

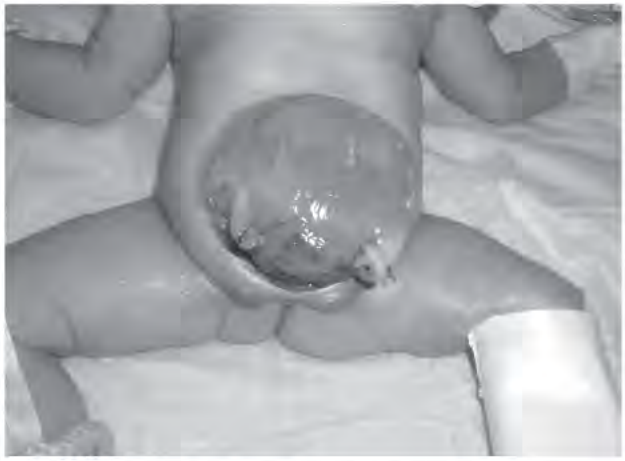

Fig. 1. General view of a newborn before surgeries with the dominating giant omphalocele

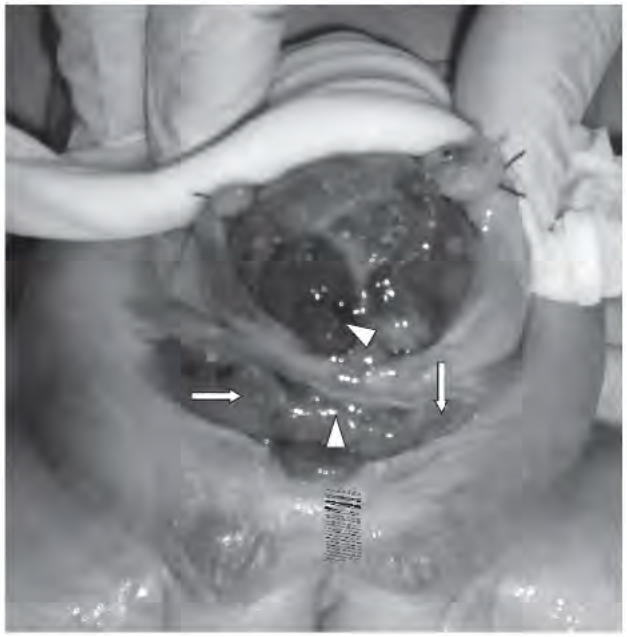

Fig. 2. Exstrophy elements before surgery incorporating intestinal extrophy in the middle (arrowheads) and bladder halves laterally (white arrows) with partially ruptured membrane of the omphalocele

Note epispadiac glans penis with bifid scrotum (arrow).

The first operation was performed on the second day of life. Because of a giant omphalocele containing the liver, the defect of the abdominal wall was closed by means of a vascular graft patch. The newborn underwent separation of the cecum plate from the bladder halves. It was tabularized lengthening the small bowel and put as a stoma. The hindgut was extremely short, located in the pelvis, and was put as a separate stoma in the left lower quadrant of the abdominal wall because of vascular compromise. The bladder halves were sewn together, and the symphysis was closed without osteotomies. Due to anuria, in spite of bilateral ureteral stents, a second surgery was performed next day and the release of symphyseal closure resulted in restoration of diuresis.

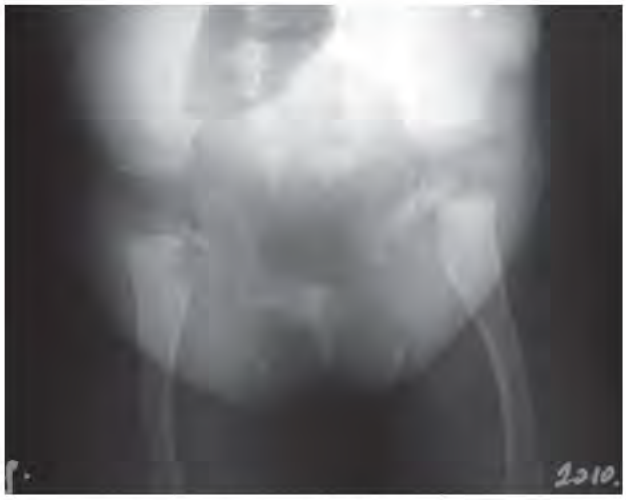

Fig. 3. Pubic diastasis before osteotomy

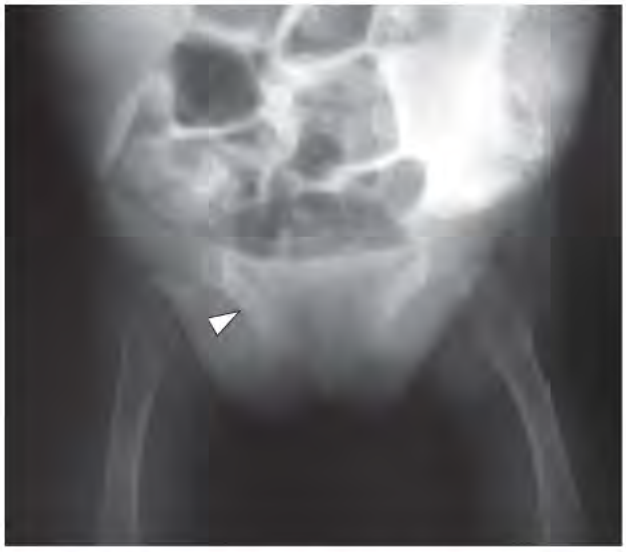

Fig. 4. One month after the repair with osteotomies

Note the improved position of the femurs and geometry of the superior pubic rami (arrowhead), lower abdominal and inguinal soft tissue. The symphyseal diastasis remains.

After a prolonged period of wound healing and stabilization of bowel transit, the newborn was discharged for gastroenterological care in the regional hospital.

Four months later, he was operated on for the third time. The weight of the infant was still very low. His weight was 3820 g. Besides this, he had huge bilateral inguinal hernias and fistula in the graft area. The graft was removed, and multiple adhesions of the small bowel were divided. The hindgut segment was found to be hypoplastic in the left iliac fossa. Because of extended duration of operation, it was not mobilized this time, planning for a different procedure to lengthen the bowel. Closure of the pubic symphysis was achieved with a help of bilateral anterior oblique mid-innominate osteotomy (Figs. 3 and 4), and the additional bladder closure was ac-

complished. Bilateral orchidopexy and hernia repair were performed during the same procedure.

The postoperative period was complicated by premature extubation and necessitating resuscitation, complicated by pneumonitis and sepsis. Complex treatment, including tracheostomy, in the intensive care unit resulted in recovery, primary wound healing, and taking off the tracheostomy tube. The child was immobilized initially with a hip spica cast, later changed to Bryant's traction for 6 weeks altogether.

There was also a problem in obtaining good fecal consistency; the infant did not gain weight. Not only because of malabsorption caused by the absence of the colon, this was also linked to intestinal allergy to cow's milk. Food intake was improved with a hypoallergenic mixture, nutritionally complete powdered formula, containing 100% free amino acids and manufactured in a milk protein-free environment together with porridge of boiled rice and mixture without lactose. It resulted in the improvement of fecal consistency, and body weight increased to 4300 g in one month.

Despite this achievement, child's growth was complicated by frequent diarrheas and malabsorption. The mother was taught to dilate the distal reconstructed blind segment of the colon by antegrade enemas. The volume of infused isotonic solution was gradually increased from 5 to 25 mL. At the age of 1 year, he was operated on again: the colonic remnant was connected to the ileum and put as a new stoma. It significantly improved fecal consistency and growth of the infant.

At the age of 1.5 years, 6 months after the last operation, the child's weight was more than 9 kg. The parents are confident in stoma care, and the child is regularly followed up by a pediatric gastroenterologist, urologist, and neurosurgeon. He is urinary incontinent with epispadias, but has no recurrent urinary tract infections or dilatation of the renal collecting system.

Discussion

Exstrophy of the cloaca was first described in 1709 by Littre and later in 1812 by Meckel. The surgical correction was introduced with the report by Rickman in 1960, describing a 3-stage procedure carried out within 8 months. The survival rate of 50% was acceptable, and male-to-female gender reassignment was recommended (1).

Cloacal exstrophy is difficult to define because the combination of anomalies is different from case to case. Severe anomaly along a spectrum that includes both epispadias and classic bladder exstrophy collectively has been termed the exstrophy-epispadias complex (EEC) (1, 2). Cloacal exstrophy has also been referred to as the OEIS complex: omphalocele, exstrophy, imperforate anus, and spinal defect (3). Beside these classic defects, most affected infants also have other anatomic anomalies (4).

It occurs probably in 1 per 200 000–400 000 live births (5). The European Society for Paediatric Urology conducted a prospective study on the incidence of bladder/cloacal exstrophy and epispadias, but the true incidence is still not clear. In Lithuania, cloacal exstrophy may occur approximately once in 10 years, but 2 cases were registered in 2 consecutive years.

In our case, only omphalocele was suspected prenatally. Nowadays, cloacal exstrophy can often be diagnosed using antenatal ultrasound. The ultrasound criteria involve no visualization of the bladder, a large midline infraumbilical anterior wall defect or a cystic anterior wall structure, an omphalocele, and lumbosacral anomalies. The antenatal diagnosis of cloacal exstrophy may cause parents to have antenatal consultation with a pediatric urologist or a surgeon and facilitate appropriate referrals before birth (6).

The classic cloacal exstrophy manifests itself as two exstrophied bladder halves divided by a strip of the exstrophied cecum, generally accompanied by a prolapsed ileal segment. In our case, there was no typical prolapse of the small bowel, so-called elephant trunk deformity. There were duplicated appendiceal orifices on the central strip. The most inferior orifice, representing the distal colon, was not duplicated in our case. It is not always evident on inspection, and the true intestinal anatomy in every case is established only during surgical exploration (Fig. 2).

Male genitalia are typically represented as an epispadiac penis on widely separated pubic bones. In female fetuses, the Mullerian duct orifices may be exstrophied below the bladder mucosa, and a duplicate vagina and bifid clitoris may be present. In our case, it was difficult for the obstetrician to determine the sex of the newborn from external examination. Male sex was confirmed in a tertiary center by a specialist after examination of external genitalia and by karyotype investigation.

Historically, a genetically male infant with cloacal exstrophy and a phallus of inadequate size for reconstruction was often assigned to female gender, performing early orchidectomy with subsequent hormone replacement at puberty. This concept was accepted, given the prevailing opinion on gender neutrality in the newborn and importance of sex of rearing at that time. The appropriateness of this decision has now been re-evaluated by several authors, leading to increased awareness but also additional controversy in the gender reassignment of these patients (4). It is important to remember that cloacal exstrophy is not a disorder of sexual differentiation, and those genetically male patients have histologically normal testicles with a normal response to antenatal androgens in the utero, as would be expected (7). The short penis and marked chordae is attributed to abnormally separated corporal bodies on the ischiopubic rami. Reconstructive procedures

for epispadias have recently improved, but still carry a high risk of complications, remaining short penis, and retrograde, if any, ejaculation. In females, after exstrophy reconstruction, the perineum seems to be placed anterosuperiorly, the vagina could be duplicated and short, with a high risk of uterine prolapse during or after puberty. In spite of various techniques, the final cosmetic and functional outcome also remains to be improved (8).

Surgery performed to repair cloacal exstrophy is usually carried out in the neonatal period, during the first 72 h if possible, in collaboration of pediatric surgeons and urologists. Neurosurgical consultation in terms of accompanying spinal dysraphism should be held as soon as the infant is medically stable. Early correction diminishes bacterial colonization of exposed viscera and may reduce the need for pelvic osteotomy. An individualized approach toward reconstruction of the genitourinary and gastrointestinal tract, whether in a single or multistage procedure, is commonly acknowledged as producing the best long-term outcomes. The objectives of management include secure abdominal wall and bladder closure, preservation of renal function, prevention of short bowel syndrome, reconstruction of genitalia adequate functionally and cosmetically, and achievement of acceptable urinary and fecal continence (7). During the third surgery, after assessing the child's age, low weight, great degree of symphyseal diastasis, immaturity of bone tissue, and other associated pathologies, it was decided to perform a bilateral anterior oblique mid-innominate osteotomy, which seems to give the optimum mobility and reduced hemorrhage risk as compared to, for example, posterior osteotomy. In our case, it allowed the rotation of pubic bones and improved the bladder closure (Fig. 4).

The objective of early treatment should be to utilize an intestine for nutrition and to functionalize the hindgut so that it could develop. For these reasons, it is recommended that the cecum, which easily could be left as a natural bladder augmentation, should be separated from the bladder, closed, and functionalized with the hindgut. If the cecal closure is tenuous, it should be protected with a temporary ileostomy that can be closed in a few weeks. The hindgut also should not be used for reconstruction of the urinary tract. As this short piece of bowel grows, it will develop water absorptive capacity that makes a pull-through or a colostomy much more manageable (3). We managed to preserve the hindgut and tubularized cecum after initially leaving it defunctionalized because of small size and doubtful blood supply. In our opinion, progressive dilatation by antegrade enemas of isotonic solution was an important factor for the success of subsequent bowel lengthening.

In the first few years of life, emphasis should be placed on growth and development of these children. The urinary tract should be left in a condition that drains freely so that the baby does not have recurrent urinary tract infections. Patient growth and nutrition needs to be monitored carefully, and regular renal ultrasound scans should be obtained to assess renal growth (9). The next reconstruction (epispadias and bladder neck repair) will be planned depending on an increasing bladder volume at the age 3 to 5 years.

Cloacal exstrophy remains a rare and challenging diagnosis and must be managed only in tertiary centers with all necessary specialists and units. In case of a small country, training of specialists in referral centers is of paramount importance. The optimal treatment of cloacal exstrophy must address many different aspects of an individual, from the timing and type of repair to genital reconstruction and quality-of-life issues (10). Advances in medical and surgical management have allowed for dramatically improved survival and continence rates, but even with the best care, these children will require lifelong support.

Statement of Conflict of Interest

The authors state no conflict of interest.

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