A previously unreported variant of exstrophy cloaca

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Cloacal exstrophy, a rare and complex congenital anomaly, presents with omphalocele; exstrophied bilateral hemibladders with ureteric or müllerian remnant orifices; central exstrophied ileocecal bowel plate with superior orifice of the terminal ileum, inferiorly, the colon, and centrally, the appendix; bifid rudimentary external genitalia; separated pubic rami; low-set umbilicus; and epispadias in the classic form. A newborn case of cloacal exstrophy presenting without an exstrophied intestine and vesicointestinal fistula is reported. The clinicopathologic features of this previously unreported variant of cloacal exstrophy are discussed with special emphasis on embryologic basis. Exstrophied bowel is the main component of exstrophy cloaca, which makes our case unique with regard to the absence of exstrophied bowel and vesicointestinal fistula. This well-known fact is not applicable to the present case. We think that some other unknown mechanisms must be at work for the development of the cloacal exstrophic anomaly presenting with a shortened intact colon ending with an anteriorly located anus. Normal development of the hindgut primarily depends on the normal formation of the cloacal membrane. The basic morphogenetic processes that consist of cell deposition, fusion, and merging should achieve the precise balance between cell proliferation and apoptotic cell death both in hindgut and cloacal membrane development. Unsatisfactory explanations of many similar malformations are primarily due to the lack of accurate and illustrative findings in different fields of embryology. The present case confirms that further studies are required to clarify the various theories in order to achieve more satisfactory explanations for these types of rare anomalies.

Key words: cloacal exstrophy, variant.

Cloacal exstrophy, a rare and complex congenital anomaly, presents with omphalocele; exstrophied bilateral hemibladders with ureteric or müllerian remnant orifices; central exstrophied ileocecal bowel plate with superior orifice of the terminal ileum, inferiorly, the colon, and centrally, the appendix; bifid rudimentary external genitalia; separated pubic rami; low-set umbilicus; and epispadias in the classic form[1]. Anomalies of other systems frequently accompany this abnormal embryogenesis.

The exstrophy variants spectrum consists of mainly pseudoexstrophy, duplicate exstrophy, superior vesical fistula, superior vesical fissure, and covered exstrophy with visceral sequestration. These variants have the musculoskeletal defect of a widely separated pubis and divergent recti, but they differ from classical form in that the bladder has varying skin cover with most probably an intact sphincter mechanism and urethra and less associated life-threatening anomalies, with a better prognosis. Although the presenting defects of the exstrophy variants are well described, the variations, multiplicity, and modifications of embryological pathogenesis can not be distinguished from each other by clear cut features and characteristics[2]. The current case is unique with regard to the absence of exstrophied intestine and vesicointestinal fistula and has raised questions about the well-known pathogenesis of this malformation. The aim of this report was to discuss the clinicopathologic features of this previously unreported entity with special emphasis on embryologic pathogenesis.

Case Report

A male newborn was referred to our hospital on his first day of life with the presumptive diagnosis of exstrophy cloaca. He was born to a 31-year-old woman after a 38-week uncomplicated pregnancy by normal spontaneous vaginal delivery. The parents were nonconsanguineous and healthy with no history of congenital anomalies. No prenatal, maternal, or fetal investigations had been performed.

On admission, his length and weight were at the 50th percentile, blood pressure was 80/50 mmHg, axillary temperature was 37°C and pulse was 110/bpm. Physical examination revealed an omphalocele sac containing liver and small intestine, exstrophied bilateral hemibladders without any kind of exstrophied strip of intestine, anteriorly located anal orifice, bifid penis, widely separated pubic bones, and bilateral undescended testis accompanied by inguinal hernia.

Laboratory investigations showed normal findings with regard to complete blood count and liver and renal function tests. Bilateral moderate hydronephrosis and patent ductus arteriosus were detected on ultrasonography and echocardiography, respectively.

After appropriate preoperative preparation, the surgical intervention was performed. The omphalocele sac was separated from the liver and intestines and excised. The small intestines and cecum were normal. There was a shortened colon ending with an anteriorly located wider anal orifice through the posterior surface of the bladder halves (Fig. 1). Posterior wall of the bladder was adherent to the anterior wall of
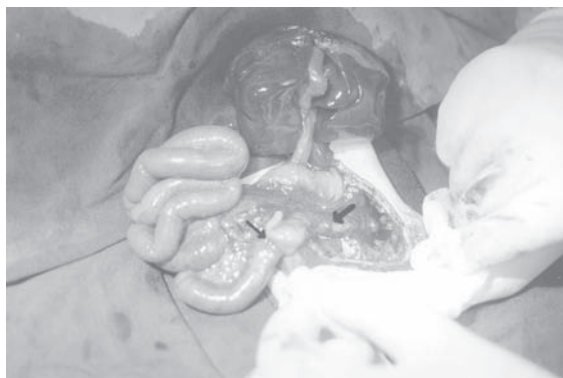

Fig. 1. Ileocecal valve (arrow), shortened colon (bold arrow) and intact rectum reaching the perineum behind the lifted bladder halves.

the intact rectum (Fig. 2). The hemibladders were excised from the rectum and primary bladder closure was done after catheterization of ureters (Fig. 3). Bladder neck was formed

Fig. 2. Posterior wall of hemibladders (arrow) are strictly adherent to the anterior wall of the rectum.

Fig. 3. Posterior wall of hemibladder was dissected free from the anterior surface of the rectum (arrow).

and tightened by paraureteral muscle and fascia flaps after pubis approximation was performed by orthopedic surgeons. Ureteroplasty was performed over 8F urethral catheter (Fig. 4). Bilateral inguinal hernia repair was also performed. All intestines were placed into the abdomen. Primary repair of abdominal wall was achieved using a 15x10 cm propylene mesh due to enlarged liver in order to avoid intraabdominal pressure increase.

During the postoperative period, intraabdominal pressure monitoring was normal and the patient was extubated on the second day. However, sepsis occurring on the sixth day was complicated with disseminated intravascular coagulopathy and the patient died at the end of the first postoperative week.
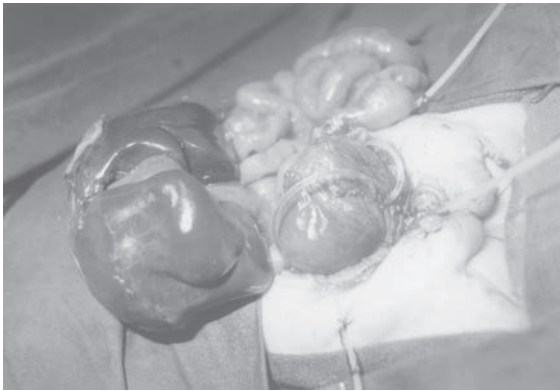
Fig. 4. Primary bladder closure with pubis approximation and ureteroplasty.

Discussion

Cloacal exstrophy occurs once in 200,000 to 400,000 births. In spite of a long tradition of embryological research, the abnormal development process leading to cloacal exstrophy is still speculative. Theories about the pathogenesis include abnormal persistence and abnormal perforation of the cloacal membrane, an oversized cloacal membrane, insufficient bilateral migration of mesodermal cells originating the primitive streak, resulting in deficient fusion and subsequent median disruption of the abdominal and bladder wall, and an oversized cloacal membrane subdivided into an infraumbilical and definitive cloacal membrane by fusion of both swellings of the genital tubercle[3-6].

It is generally accepted that normal development of cloaca into the dorsal anorectum and the ventral urogenital tract depends on the proper subdivision of the cloaca by the so-called urorectal septum[7]. However, there is no consensus among the investigators about the nature and development of the urorectal septum, which has a major role in cloacal anomalies. It was proposed that the cranial part of the septum grows downward while in the caudal part, lateral ridges fuse to form the septum in this area[8]. In contrast, the major role of the urorectal septum in cloacal differentiation has been denied and it was proposed that shift of caudal cloaca and migration of the rectum play a major role in establishing the anal opening and development of the hindgut[9].

On the other hand, according to recent studies, it is more likely that a normal urorectal septum is the result of normal cloacal development rather than its cause, as believed previously[7].

Based on this assumption, it is reported that a defective cloacal anlage results in a missing or misplaced anal orifice and abnormal communication between the rectum and the ventral urogenital tract[3,10.] Additionally, dorsal cloacal membrane and the dorsal cloaca have been found to be missing in exstrophic anomalies rather than overdevelopment as proposed previously[11]. According to some researchers, fusion between the umbilical ring and the cloacal membrane does not take place in the median of the abdominal wall. Moreover, the mesoderm of the abdominal wall and external genitalia originate not only from the primitive streak but also from the body wall, which brings new insights into abdominal wall defects[6,12].

It is widely believed that if the cloacal membrane ruptures before the urorectal septum descends (5-week human embryo), exstrophy of a central bowel field flanked by exposed hemibladder mucosa results. If cloacal separation begins by the downgrowth of the urorectal septum (6-week human embryo) and then cloacal membrane disintegration occurs, the exstrophied gut may lie caudad to a single exstrophied bladder. Rupture of cloacal membrane in the 7th week results in classic bladder exstrophy only[13]. In light of these findings, it is obvious that exstrophied bowel is the main component of extrophia cloaca, which makes our case unique with regard to the absence of exstrophied bowel and vesicointestinal fistula. This well-known theory is not applicable in the present case. We think that some other unknown mechanisms must be at work for the development of cloacal exstrophic anomaly presenting with a shortened intact colon ending with an anteriorly located anus. Normal development of the hindgut primarily depends on the normal formation of the cloacal membrane. The basic morphogenetic processes that consist of cell deposition, fusion, and merging should achieve the precise balance between cell proliferation and apoptotic cell death both in hindgut and cloacal membrane development[14,15].

Based on the above-mentioned embryologic pathogenesis, it is clear that "unknown" is still more than "known" or in other words "beliefs". None of the mentioned theories explain the pathologic morphogenesis in the present case. Unsatisfactory explanations of many similar malformations are primarily due to the lack of

accurate and illustrative findings in different fields of embryology. Therefore, we, pediatric surgeons, are highly confused when trying to understand the embryologic backgrounds of uncommon malformations. The present case confirms that further studies are required to clarify the various theories in order to achieve more satisfactory explanations for these types of rare anomalies.

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