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Case report

Congenital Malformation: Bladder Exstrophy with Anal Atresia

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Abstract

It is an infrequent congenital pathology in pediatrics when children are born with an open malformation. Bladder exstrophy is a rare congenital pathology manifests itself as isolated defect or as part of several defects as a complex.

We present a clinical case of nine-month-old baby girl suffered from absence of anterior abdominal wall and anterior bladder wall, divergence of bones pelvic articulation and absence of anus. The congenital defect was detected intrauterine by ultrasound at the third screening. After birth, an operation with a colostomy was performed. Due to the severe diagnosis, a decision was made to hospitalize her at our medical facility for further examination and treatment.

Key words: *bladder exstrophy, anus atresia, bladder exstrophy-epispadias complex, plastic surgery, osteotomy.*

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Introduction

The bladder exstrophy-epispadias complex (BEEC) is an anterior midline defect with variable expression comprising a spectrum of defects involving the abdominal wall, pelvis, urinary tract, genitalia, and occasionally the spine and anus [1]. The incidence of bladder exstrophy has been reported as varying from 1 in 20.000 to 30.000 live births, with a male: female ratio of 2-3:1 [2]. Epispadias is generally diagnosed at birth, although its presentation is dependent on severity and sex. It consists of a dorsal located ectopic urethral meatus as a result of non-closure of the urethral plate during embryological development [3]. Surgical treatment of classical bladder exstrophy not only involves the bladder closure, but it also includes procedures such as epispadias repair, bladder neck reconstruction, bladder augmentation, and ureteric reimplantation. Additionally, the abnormalities in the bony pelvis and the pelvic floor need to be addressed along with exstrophy repair [4].

In our case we want to present a rare congenital embryological defect as bladder exstrophy not as isolated pathology, but as a complex with anal atresia which is amenable to surgical treatment.

Case Report

Nine-month-old baby girl was admitted to our hospital due to a physical ailment. Congenital defect was detected firstly intrauterine on ultrasound third screening at 30 weeks 6 days where congenital malformation of the urogenital system, omphalocele was observed. Further, at 40.5 weeks cephalic presentation, double umbilical cord entanglement around the fetal neck were detected at the ultrasound. After giving birth, child of undetermined sex, weight at birth - 3460 g, height - 54 cm, on Apgar scale - 8-9. The patient was immediately taken for surgery "Colostomy removal" because of anal atresia.

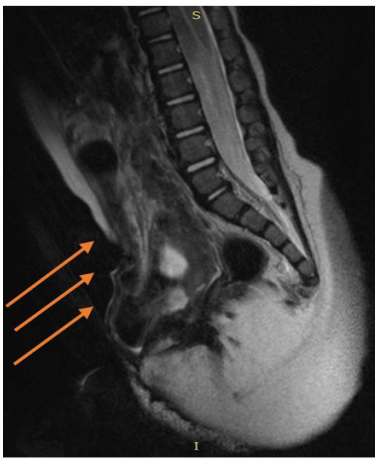

Figure 1 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: Defect of the anterior abdominal wall (orange arrows). Urinary bladder is displaced and located on the anterior abdominal wall.
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). Sagittal MRI, T2-tse-sag-p2, time to echo (TE) 112 ms, repetition time (TR) 6771.6 ms

After five moths there was a consultation with a pediatric urologist in our clinic with the diagnosis: Cloacal bladder exstrophy, recommended hospitalization in the department of urology and kidney transplantation by portal for further examination and surgical treatment.

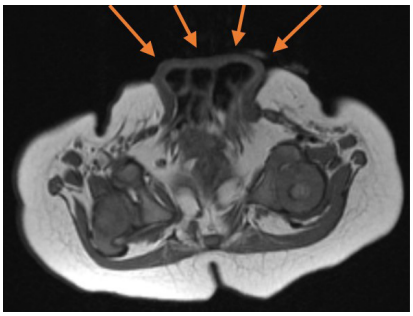

Figure 2 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: Defect of the anterior abdominal wall, urinary bladder is displaced and located on the anterior abdominal wall, the posterior wall of the urinary bladder is undifferentiated (orange arrows)
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). Axial MRI, T1-tse-tra-lymph-nodes, time to echo (TE) 12 ms, repetition time (TR) 731 ms

The first thing she undergoes is a pelvic magnetic resonance tomography (MRI), where at the level of the urinary bladder there is a 51 mm defect of the anterior abdominal wall (Figure1), through this defect the

urinary bladder is displaced and located on the anterior abdominal wall, the posterior wall of the urinary bladder is undifferentiated, there are bowel loops in the cavity with a small amount of free fluid on the lower contour (Figure 2, 3).

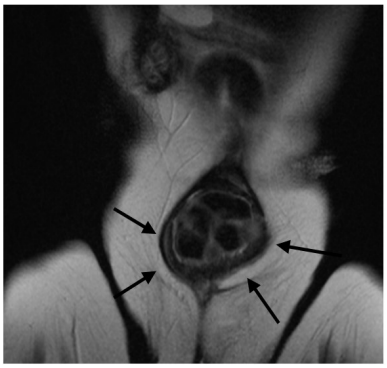

Figure 3 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: Bowel loops in the urinal bladder cavity with a small amount of free fluid on the lower contour (black arrows)
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). Coronal MRI, T2-tse-cor-p2, time to echo (TE) 112 ms, repetition time (TR) 6000 ms

The long bones were not fully developed, there was a diastasis of up to 46 mm between them (Figure 4). The anal part of the rectum is undifferentiated, the rectal ampulla is in a typical location, filled with air (Figure 5). A stoma was detected in the anterior abdominal wall on the right side (Figure 6,7).

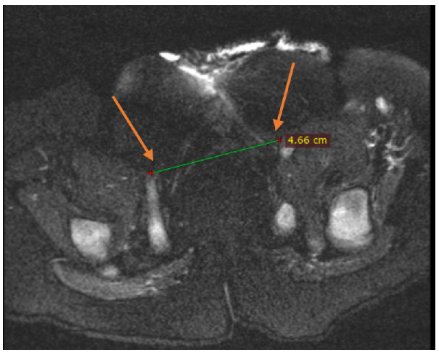

Figure 4 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy
FINDINGS: Long bones were not fully developed, diastasis of up to 46 mm between them (orange arrows).
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). Axial MRI, T2-tse-axi-fs-p2, time to (TE) 1.8 ms, repetition time (TR) 4.5 ms

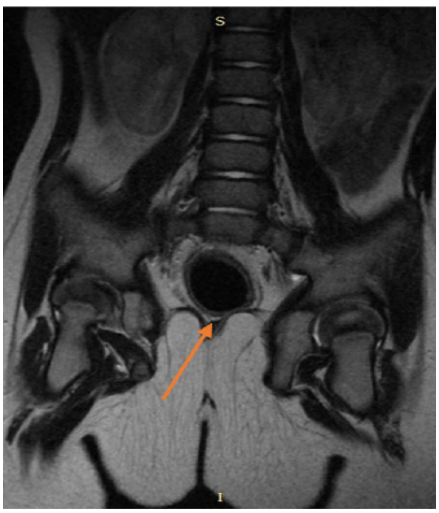

Figure 5 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy
FINDINGS: Anal part of the rectum is undifferentiated, the rectal ampulla is in a typical location, filled with air (orange arrow).
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). Coronal MRI, T2-tse-cor-p2, time to (TE) 112 ms, repetition time (TR) 6000 ms

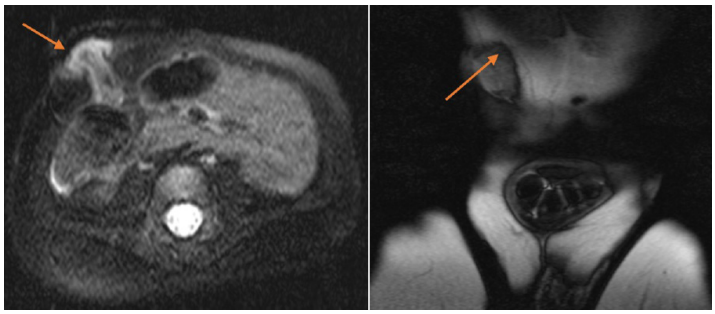

Figure 6, 7 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: Stoma in the anterior abdominal wall on the right side (orange arrows).
TECHNIQUE: Magnetic resonance, 1.5 T MR System (MAGNETOM Avanto, Siemens Healthcare, Germany). coronal MRI, T2-tse-fs-tra-mbh-pat2, T2-tse-cor-p2, time to (TE) 106 and 112 ms, repetition time (TR) 4000 and 7632 ms

Based on these findings, she was diagnosed with abnormalities of the urinary tract: bladder ectrophy with anal atresia and pelvic articulation bone discrepancy.

After all diagnostic examinations patient was offered surgical correction of the defect.

Discussion

The first description of bladder exstrophy was noted on Assyrian tablets nearly 4000 years ago [5]. Bladder exstrophy-epispadias complex develops at around 4 to 6 weeks of gestation, when the cloacal membrane is prematurely invaded by mesoderm to build the abdominal wall for the genitourinary system. Timing of the rupture may determine severity. If the cloacal membrane ruptures before 4 weeks of gestation, cloacal exstrophy results, and if it ruptures after the urorectal septum has descended at 6 weeks, epispadias or bladder exstrophy results [6].

Bladder exstrophy is a rare congenital malformation of the genitourinary system with an estimated incidence of approximately one per 50.000 live births [7]. Patients with the exstrophy-epispadias complex have a widened pubic symphysis due to malrotation of the pelvic innominate bones. Although, the most profound anomaly is the defect in the ventral abdominal wall fascia, resulting in an open and anterior positioned bladder and urethra [8]. Bladder exstrophy can be firstly diagnosed during prenatal development with fetal ultrasonography between the 15th and 32nd week of pregnancy [9]. With the growing understanding of the detrimental effects of radiation in children, MRI is progressively been utilized in the preoperative work-up and post-surgical follow-up of these patients [10]. Despite several modifications over the years, currently there are two main approaches to bladder exstrophy repair: modern staged repair of exstrophy (MSRE) and complete primary repair of exstrophy (CPRE) [11]. After surgery urinary diversion in some series seems to result in better ejaculatory hence fertility outcome. Male exstrophy patients are reported to have more of psychosexual disturbances as compared to the females. Most of the female patients have normal fertility but contrary to this most of their counterpart male patients have significantly suboptimal fertility [12]. Despite the statistics, the incidence of the defect prevails in boys, in our case there was a girl who had this genitourinary malformation, which does not exclude the rarity of our case. The defect was detected in utero, which suggests that ultrasound is the first choice to diagnose complex malformations. Also, due to atresia of the anus, the girl was immediately operated on after birth and a colostomy was taken out. Due to the fact that the patient was constantly monitored by the pediatrician and surgeon at the place of residence in her city, the surgery for suturing and closure of the defect was performed 9 months after delivery at our clinic.

Treatment. The indication for surgery was the absence of the anterior bladder wall, anterior abdominal wall, and symphysis divergence. In the pre-operative period, the day before the operation, the patient underwent X-rays of the hip joints to clarify the degree of divergence of the pubic symphysis (Figure 8).

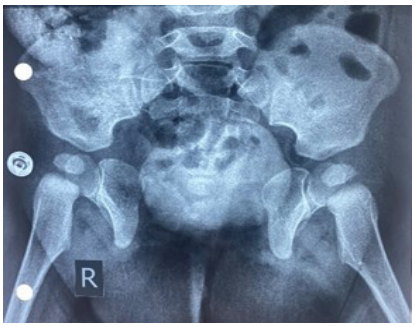

Figure 8 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: An x-ray of the pelvic bones and hip joints in straight projection shows a divergence of up to 4.1 cm in the pubic pelvic bones, which corresponds to a grade 3 symphysis divergence.
TECHNIQUE: X-ray (Siemens Multix Top, Germany). Effective equivalent dose (EED) is 0,020 millisievert

Our patient was offered the suggested scope of surgery, which was bladder plastic surgery, bladder neck surgery, urethrovaginoscopy, and osteotomy on both sides. Bladder neck reconstruction is typically performed when the child is ready to be dry and is able to cooperate with a voiding program. Most patients undergo this procedure at age 4 to 5 years of age [13]. The operation was divided into two processes. The first was a bladder exstrophy repair vaginoplasty. Bladder plasty was performed using local tissue with the formation of a bladder neck. Further, orthopaedists performed an osteotomy of the iliac bone on both sides and installed metal fixators (Figure 9).

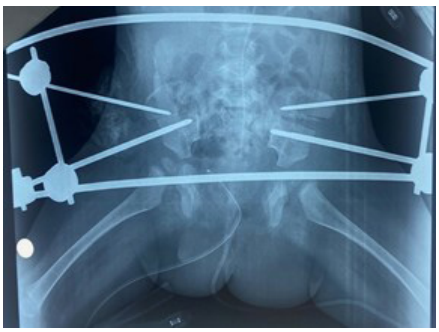

Figure 9 - A nine-month-old baby girl diagnosed with congenital malformation: bladder exstrophy with anal atresia and pelvic articulation bone discrepancy

FINDINGS: Post-operative condition - pelvic osteotomy on both sides according to Salter. Metallosynthesis. There is a discrepancy of up to 4.1 cm in the pubic bone of the pelvis.
TECHNIQUE: X-ray (Siemens Multix Top, Germany). Effective equivalent dose (EED) is 0,020 millisievert

The postoperative wound was repaired. A neo-urethra on a urethral catheter was fostered. Nodular sutures to the skin were applied, also an aseptic dressing. A urethral catheter was placed in the bladder. There were no post-operative complications.

Surgical treatment in the complex of bladder oestrophy and epispadias of the genitals and cloaca is the best method. Also, a staged approach to urogenital reconstruction gives excellent treatment results when both paediatric surgeons, orthopaedists and anaesthetists are involved. All of this together enables young patients to live under normal natural conditions.

Conclusions

In the diagnosis of severe bladder malformations such as exstrophy with genital epispadias, radiodiagnosis takes precedence. In combination with ultrasound, radiography and MRI, we can make the right diagnosis, suggest the exact course of surgery, help the patient with treatment and prolong his or her life. Post-operative follow-up of the patient is also important to us, which also requires instrumental radiological diagnostics primarily to rule out complications.

Ethical aspects. The patient's legal representatives gave informed consent to the use of medical information and photographs.

Conflict of interest. No conflict of interest has been declared.

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Туа біткен ақау: анальды атрезиямен қуық экстрофиясы

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Түйіндеме

Қуық экстрофиясы - оқшауланған ақау ретінде немесе кешендегі бірнеше ақаулардың бөлігі ретінде көрінетін сирек туа біткен патология. Бұл педиатрияда сирек кездесетін туа біткен патология, онда балалар ашық ақаумен туылады.

Біз іштің алдыңғы қабырғасы мен қуықтың алдыңғы қабырғасының жетіспеушілігінен, жамбас сүйектерінің алшақтығынан және анустың жетіспеушілігінен зардап шеккен 9 айлық қыз баланың клиникалық жағдайын ұсынамыз. Туа біткен ақау үшінші скринингте ультрадыбыстық зерттеу арқылы жатырда анықталды. Босанғаннан кейін колостомиямен операция жасалды. Ауыр диагнозға байланысты оны әрі қарай тексеру және емдеу үшін біздің медициналық мекемеге жатқызу туралы шешім қабылданды.

Түйін сөздер: *қуық экстрофиясы, анальды атрезия, қуық экстрофиясы-эписпадия кешені, пластикалық хирургия, остеотомия.*

Врожденный порок развития: экстрофия мочевого пузыря с атрезией заднего прохода

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Резюме

Экстрофия мочевого пузыря - редкая врожденная патология, проявляющаяся как изолированный дефект или как часть нескольких дефектов в комплексе. Это нечастая врожденная патология в педиатрии, когда дети рождаются с открытым пороком развития.

Мы представляем клинический случай девятимесячной девочки, страдавшей отсутствием передней брюшной стенки и передней стенки мочевого пузыря, расхождением костей тазового сочленения и отсутствием заднего прохода. Врожденный дефект был обнаружен внутриутробно с помощью ультразвукового исследования при третьем скрининге. После родов была проведена операция с колостомией. В связи с тяжелым диагнозом было принято решение госпитализировать ее в наше медицинское учреждение для дальнейшего обследования и лечения.

Ключевые слова*: экстрофия мочевого пузыря, атрезия заднего прохода, экстрофия мочевого пузыря - комплекс эписпадии, пластическая хирургия, остеотомия.*