PREGNANCY WITH CONGENITAL ABNORMALITIES FROM MARRIAGE OF A DIFFERENT ETHNIC: A VERY RARE CASE

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Abstract

Bladder exstrophy (BE) is one of the rare congenital abnormalities. This congenital disorder is also serious, with characteristics such as a spectrum of disorders involving the walls of the ventral body, the urogenital tract, the pelvis, the spine, and the anus. The incidence rate of this case is 1 per 30,000 to 50,000 live births. The case study reported the findings of congenital bladder extrophy in a 29-year-old Acehnese woman married to an Indian citizen, with a diagnosis of G3P0A2 38-39 weeks pregnant, coming with complaints. The mother felt the discharge of water. The patient admitted to being 9 months pregnant with HPHT 27/1/2021 and TTP 3/11/21 according to the gestational age of 38-39 weeks. The patient has a history of the disease with type 2 diabetes mellitus. Initial laboratory examination findings showed hyperglycemia at 256 mg/dL. Ultrasound examination found a single head presentation of the fetus alive with a mass in the abdomen of the fetus. The patient is planning to have an emergency cesarean section and is consulted by the internal medicine department for blood sugar regulation. Postpartum findings in infants with hernia, *umbilical cord*, *Bladder Exstrophy*, epispadia, and symphysiolysis will be consulted to the Division of Pediatric Surgery. This case report describes a pregnancy with congenital abnormalities and *Bladder Exstrophy*. Prenatal diagnosis allows the medical team to prepare for a multispecialty approach. It also gives parents time to prepare and adjust to expectations related to labor and the neonatal period, as well as potential complications and long-term prognosis.

Keywords: Bladder exstrophy, rare congenital abnormalities, umbilical cord

INTRODUCTION

Bladder exstrophy (BE) is a rare congenital disorder that falls under the spectrum of disorders *bladder-exstrophy-epispadias complex* (BEEC) (Appeadu-Mensah et al., 2020). This congenital disorder is also serious, characterized by a spectrum of abnormalities involving the walls of the ventral body, urinary tract, genitals, pelvic bones, spine, and anus.(Anand & Lotfollahzadeh, 2021; Beaman et al., 2021) *Bladder exstrophy* It is estimated to occur at 3.3 per 100,000 live births and is dominated by the male sex, while other publications state an incidence rate of 1 per 30,000 to 50,000 live births.(Anand & Lotfollahzadeh, 2021; Beaman et al., 2021; Joshi et al., 2018; Lee & Shim, 2003)

Various theories about etiopathogenesis have been put forward. Marshall and Muecke's theory is the most widely accepted. Growth abnormalities in the cloaca membrane cause *a wedge effect* and prevent medial migration of mesenchymal tissue between the ectodermal and endodermal layers on the lower abdominal wall (Mafruchati, 2023). As a result, the lower abdominal wall is not well formed. The rupture of the cloacal membrane further causes herniation of all abdomens and leads to the clinical picture of BEEC. It has been stated that the *wedge effect* can also occur due to abnormalities originating from the cephalad of the genital primordia of the tuberculum genital.(Anand & Lotfollahzadeh, 2021; Beaman et al., 2021)

In genetic research, Beaman et al. proposed that the genetic variants relevant to the BEEC etiology are very heterogeneous, as shown by congenital heart abnormalities.(Beaman et al., 2021) Currently, the 22q11.2 duplicate is the genetic variant most commonly associated with BEEC.(Yerkes, 2019)

Diagnosis *Bladder Exstrophy* is clinical and can be made as soon as the baby is born in most cases (Mallmann et al., 2019). The fleshy red mass that comes out of the suprapubic region depicts an open bladder (Anand & Lotfollahzadeh, 2023). There was a continuous leak of urine from the masses. For the record, external exposure to the bladder correlates with the degree of pubic diastasis. Therefore, bladder prolapse can range from the open trigonum to the entire bladder.(Anand & Lotfollahzadeh, 2021; Beaman et al., 2021; Lee & Shim, 2003; Promm & Roesch, 2019)

Prenatal diagnosis can be made during ultrasound screening.(Promm & Roesch, 2019) When a dense mass is seen in the lower abdomen of the fetus at prenatal time, there are many differential diagnoses. But if it can be conclusively that the mass is from the bladder, the correct diagnosis will be easier to make.(Lee & Shim, 2003)

RESEARCH METHODS

Research Design

This study employs a qualitative research design with a descriptive approach. This approach was chosen to provide an in-depth understanding of the phenomenon under study, namely pregnancy with rare congenital abnormalities. The descriptive approach allows the study to explore case details comprehensively, document relevant facts, and explain the medical and social implications of the investigated case.

Research Location

The study was conducted at Dr. Zainoel Abidin Regional General Hospital (RSUD), Banda Aceh, a major referral center in Aceh. This location was selected for its advanced healthcare facilities capable of managing high-risk pregnancies and congenital abnormalities.

Research Period

The research was carried out over a specified period, from January to December 2024. This timeline included phases of observation, data collection, analysis, and the preparation of the final report.

Research Subjects

The primary subject of this study is a 29-year-old Acehnese woman with a history of high-risk pregnancy. The reported case involves a pregnancy with a very rare congenital abnormality, bladder exstrophy. In addition to the patient, the study involved medical teams, including obstetrics and gynecology specialists, pediatric specialists, and pediatric surgery teams.

Aspects Examined

The study covered various aspects, including:

1. The patient’s medical history, including prior conditions such as diabetes mellitus.
2. Results of physical and supporting examinations, such as ultrasonography and laboratory tests.
3. Medical management during pregnancy and delivery, including cesarean section procedures.
4. Postpartum conditions of both the mother and the baby.
5. Long-term implications of the congenital abnormality on the patient’s and family’s quality of life.

Research Instruments

The instruments used in this study include:

1. The patient's medical records as the primary data source.
2. Notes on laboratory and radiological examination results.
3. Interviews with the patient and medical team members to obtain direct perspectives on the case.
4. Intraoperative and postoperative reports as supporting documents.

Data Collection

Data were collected through direct observation, documentation, and structured interviews. Observations were conducted during examination, delivery, and postpartum care to record real-time details of events. Documentation involved utilizing medical records, laboratory results, and ultrasonography findings. Interviews were conducted to gather additional information not included in medical documentation.

Population and Sample

The study population comprised all patients with similar cases at Dr. Zainoel Abidin Regional General Hospital during the research period. However, this study utilized a case study approach, limiting the sample to one specific case relevant to the research objectives.

Data Analysis

Data were analyzed qualitatively using an inductive approach. The collected data were analyzed to identify patterns, themes, and relationships between various aspects of the study. The analysis results were used to construct a narrative that comprehensively describes the case and provides insights relevant to medical practitioners and researchers in related fields.

RESULTS AND DISCUSSION

Patient Identity

The female patient with the initials SB, number CM 1-28-08-51, is 29 years old, is Muslim with Acehnese ethnicity and works as a housewife (IRT).

Anamnesis

Main complaints : Water coming out of the birth canal

Current disease history:

The patient sent by SpOG came with complaints of water discharge that was felt 6 hours before entering the hospital, namely at 15.00 WIB. The water that came out was clear, and the underwear was soaked. The patient does not feel any mules, and the mother actively feels the fetal movements. The patient admitted to being 9 months pregnant with HPHT 27/1/2021 and TTP 3/11/21 according to the gestational age of 38-39 weeks. The patient was admitted to ANC every month by the obstetrician and gynecologist, and the midwife, never, with the last ultrasound, said the fetus was in good condition. The history of vaginal discharge is denied. Defecate and urinate within normal limits.

History of previous diseases: A history of diabetes mellitus exists, while hypertension, heart disease, and asthma do not exist Family history of disease: The patient's biological father suffers from Diabetes Mellitus.

Menarche History: At 14 years old, regular 6-7 days, 2-3 changes of pads, 28-day cycle, negative dysmenorrhea

Marriage History: 1 time, age 25 years old

Childbirth History:

1. Abortion, 2 weeks, curettage
2. Abortion, 4 weeks, curettage
3. Pregnant now

Family History: None

Social History: The patient denies having a habit of smoking and drinking alcohol. The patient is a housewife, and her husband is self-employed.

Physical examination

Vital Sign

Composmentis Patients. In the blood pressure examination, 119/79 mmHg, pulse rate 81 x/min, respiration 20 x/min and Axila temperature: 36.7 (Evangelidis et al., 2004)C

General Physical Examination

Head : palpebra conj inf pale (+/+), jaundice (-/-), pupil reflex (+/+) isocor, palpebra edema (-/-)

Neck

JVP : PR + 0 cmH2O

Lymph nodes : no magnification

Parotid gland & thyroid: no magnification found

Thorax

Cast: Inspection: No pulsation of the chorus ictus, spider navi (-),

Palpation: the cactus of the chorus is palpable in the ICS V 2 cm from *Midclavicular Line* sinistra, strong lift (-), *Thrill* (-)

Percussion: upper limit of the heart ICS II *Midclavicular Line* sinistra, the right border of the heart *Parasternal Line* extra, the left border of the heart is 3 cm from *Midclavicular Line* sinistra ICS V

Auscultation : S1>S2 single, regular, murmur (-)

Pulmo: Inspection : static & dynamic symmetric, retraction (-), spider nervi (-),

Palpation : Fremitus vowels (↓/N)

Percussion : sonor/sonor
dim/dim
dim/dim

Auscultation : vesicular +/+, ronkhi -/-, wheezing -/-

Abdominal:

Inspection : distention (-) spider navi (-)

Auscultation : Normal bowel noise (+)

Palpation : The liver is not palpable, the lien is not palpable, the kidneys are not palpable

Percussion : Timpani, Shifting dullness (-)

Limb : warm +/+ edema -/-

Gynaecological Status:

Inspection : Vulva/ calm urethra

Inspector : Portio livid, OUE closed, flux positive, flour negative, nitrazine test (+) pooling (+)

Vaginal touche : Posterior, soft, opening 2 cm, head in Hodge I-II.

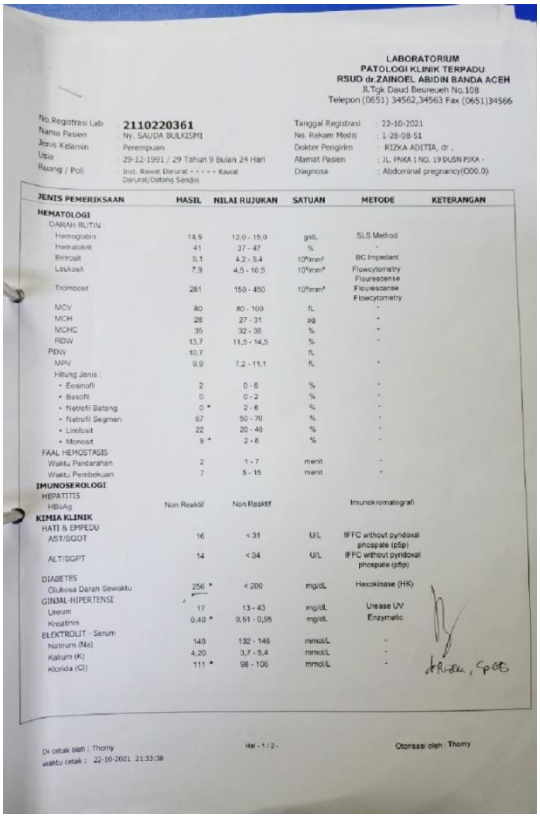
Supporting Examination

Laboratory

Hb : 14.5 g/dl
Ht : 41 %
Leu : 7,900 /mm3
Tro : 261,000 /mm3
HbsAg : Non reactive
KGDS : 256 mg/dl
your/cr : 17/0.4 mg/dl

SGOT : 16 U/L
SGPT : 14 U/L
CT : 7 minutes
BT : 2 minutes
Na : 149 mmol/L
K : 4.2 mmol/L
Cl : 111 mmol/L

The conclusion of the initial laboratory examination of this patient was obtained hyperglycemia with 256 mg/dl,



LABORATORIUM
PATOLOGI KLINIK TERPADU
RSUD dr ZAINOEL ABIDIN BANDA ACEH
Jl.Tgk Daud Beureueh No.108
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No.Registrasi Lab : 2110220361
Nama Pasien : Ny. SAUDA BULKISMI
Jenis Kelamin : Perempuan
Usia : 29-12-1991 / 29 Tahun 9 Bulan 24 Hari
Ruang / Poli : Inst. Rawat Darurat - - - - - Rawat Darurat;Datang Sendiri
Tanggal Registrasi : 22-10-2021
No. Rekam Medis : 1-28-08-51
Dokter Pengirim : RIZKA ADITTA, dr ,
Alamat Pasien : JL PKA 1 NO. 19 DUSN PJKA -
Diagnosa : Abdominal pregnancy(O00.0)

JENIS PEMERIKSAAN	HASIL	NILAI RUJUKAN	SATUAN	METODE	KETERANGAN
HEMATOLOGI					
DARAH RUTIN :					
Hemoglobin	14.6	12.0 - 15.0	gr/dL	SLS Method	
Hematokrit	41	37 - 47	%	-	
Eritrosit	5.1	4.2 - 5.4	10⁶/mm³	BC Impedant	
Leukosit	7.9	4.5 - 10.5	10³/mm³	Flowcytometry Fisuresense	
Trombosit	281	150 - 450	10³/mm³	Fleuresense Flowcytometry	
MCV	80	80 - 100	fL	-	
MCH	28	27 - 31	pg	-	
MCHC	35	32 - 36	%	-	
RDW	13.7	11,5 - 14,5	%	-	
PDW	10.7		fL		
MPV	9.9	7,2 - 11,1	fL	-	
Hitung Jenis :					
- Eosinofil	2	0 - 6	%	-	
- Basofil	0	0 - 2	%	-	
- Netrofil Batang	0 *	2 - 6	%	-	
- Netrofil Segmen	67	50 - 70	%	-	
- Limfosit	22	20 - 40	%	-	
- Monosit	9 *	2 - 8	%	-	
FAAL HEMOSTASIS					
Waktu Perdarahan	2	1 - 7	menit	-	
Waktu Pembekuan	7	5 - 15	menit	-	
IMUNOSEROLOGI					
HEPATITIS					
HBsAg	Non Reaktif	Non Reaktif		Imunokromatografi	
KIMIA KLINIK					
HATI & EMPEDU					
AST/SGOT	16	< 31	U/L	IFFC without pyridoxal phospate (p5p)	
ALT/SGPT	14	< 34	U/L	IFFC without pyridoxal phospate (p5p)	
DIABETES					
Glukosa Darah Sewaktu	256 *	< 200	mg/dL	Hexokinase (HK)	
GINJAL-HIPERTENSI					
Ureum	17	13 - 43	mg/dL	Urease UV	
Kreatinin	0,49 *	0,51 - 0,95	mg/dL	Enzymatic	
ELEKTROLIT - Serum					
Natrium (Na)	148	132 - 146	mmol/L	-	
Kalium (K)	4,20	3,7 - 5,4	mmol/L	-	
Klorida (Cl)	111 *	98 - 106	mmol/L	-	

Di cetak oleh : Thomy
waktu cetak : 22-10-2021 21:33:38
Hal - 1 / 2 -
Otorisasi oleh : Thomy

Figure 1. Patient Examination Results
Source: processed data 2024

Ultrasound

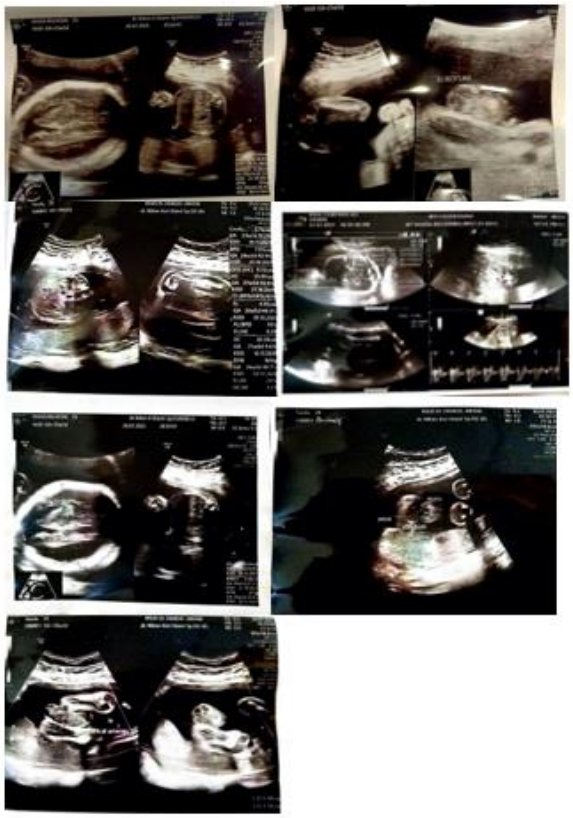


Figure 1. Patient Ultrasound Results
Source: processed data (2024)

From the ultrasound hurul, a positive FHR was obtained. The single-head presentation fetus was alive with BPD 9.28 cm, HC 33.48 cm, AC 32.57 cm, FL 6.22 cm, EFW 3119 gr, and AFI 11.9 cm.

Impressions on ultrasound biometry examination were obtained in accordance with the age of the fetus at 38-39 weeks, but a mass was also found in the abdominal region in front of the fetal symphysis with suspicion of an omphalocele.

Diagnosis

G3P0A2 38-39 weeks pregnant, JPKTH, KPD 6 hours (ICA 2), fetus with Omfalocel susp and Mother with type 2 DM

Management

a. Take care of family planning
b. SC Cito's plan for tomorrow morning
c. Consul of IPD EMD → blood sugar regulation
d. Inj Ampicillin 1 gr/6 hours

Prognosis

Quo ad Vitam: Bonam
Quo ad Sanactionam: bonam
Quo ad Functionam: bonam

A 29-year-old Acehnese woman, G3P0A2, 38-39 weeks pregnant, came to the Emergency Installation of Dr. Zainoel Abidin Banda Aceh Hospital; SpOG sent came with complaints of water discharge that was felt 6 hours before entering the hospital, namely at 15.00 WIB. The water that came out was clear, and the underwear was soaked. The patient does not feel any mules, and the mother actively feels the fetal movements. The patient admitted to being 9 months pregnant with HPHT 27/1/2021 and TTP 3/11/21 according to

the gestational age of 38-39 weeks. The patient was admitted to ANC every month by the obstetrician and gynecologist, and the midwife never, with the last ultrasound, said the fetus was in good condition. The history of vaginal discharge is denied. Defecate and urinate within normal limits. The patient had a history of the disease with type 2 diabetes mellitus.

A thorough examination was carried out at the Emergency Installation of Dr. Zainoel Abidin Banda Aceh Hospital. Initial laboratory examination findings showed hyperglycemia at 256 mg/dL. Ultrasound examination found a single-head presentation fetus alive with a mass in the abdomen. Patients are planned for emergency cesarean delivery and consulted with the internal medicine department for blood sugar regulation.

Intraoperative Findings

A pfanensteil incision was performed, and the intraoperative finding was green amniotic fluid in a small amount. The baby's head was born with a body weight of 3500 grams, a body length of 48 cm, and an Apgar score of 7/8. In babies, omphalocele appears accompanied by an atrophic blader. Next, the placenta is born completely. The abdominal wall was tightened layer by layer, and it was confirmed that there was no bleeding. The procedure is complete.

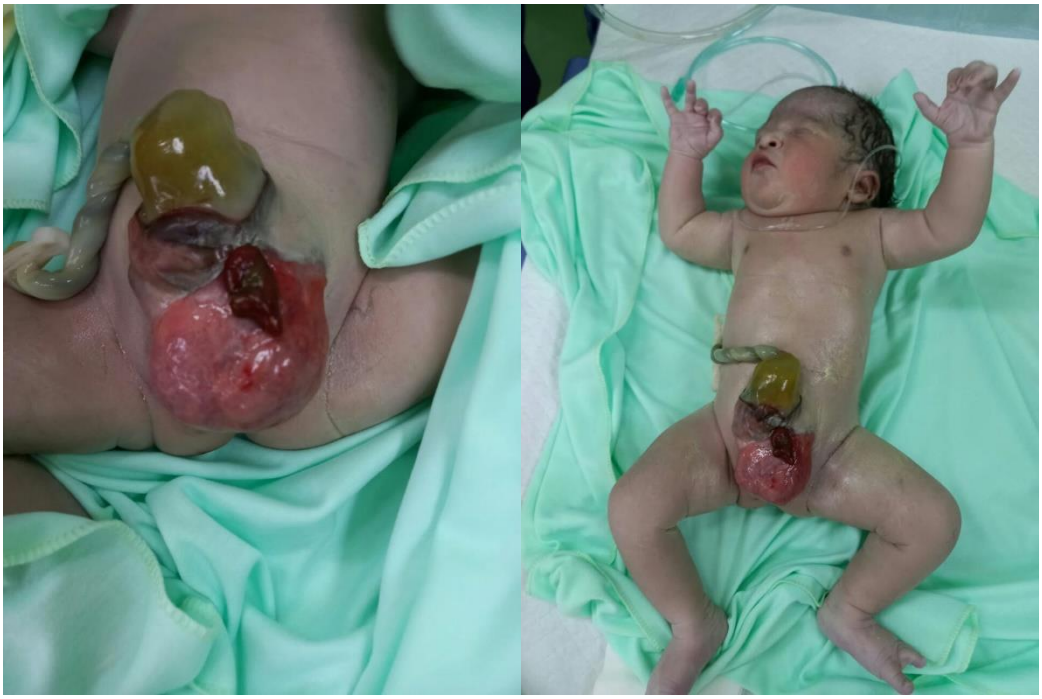


Figure 2. Infants with omphalocele and atrophied cloaca
Source: processed data (2024)

Post Operative Conditions

The patient's condition during treatment is stable.

Discussion

Bladder exstrophy is a case of congenital disorders that are very rare. The etiopathogenesis is not widely known, but Marshall and Muecke's theory explains that abnormal growth in the cloaca membrane causes *a wedge effect* and prevents medial migration of mesenchymal tissue between the ectodermal layer and the endodermal layer on the lower abdominal wall. As a result, the lower abdominal wall is not well formed. The rupture of the cloacal membrane further causes herniation of all abdomens and leads to a clinical picture.(Anand & Lotfollahzadeh, 2021; Beaman et al., 2021) Prenatal diagnosis can be confirmed during ultrasound screening.(Promm & Roesch, 2019) When a dense mass is seen in the lower abdomen of the fetus at prenatal time, there are many differential diagnoses.

But if it can be conclusively that the mass is from the bladder, the correct diagnosis will be easier to make.(Lee & Shim, 2003)

Weiss et al. in 2020 provided key findings for prenatal diagnosis, anatomical images on ultrasound, and MRI in *bladder* and *Cloacal Exstrophy*. The study involved 21 births between 2000 and 2018 in evaluating the presence of kidneys, the presence of an *Anal Dimple*, the location of the abdominal insertion of the umbilical cord relative to the defect, the number of central blood vessels, and the spinal disorder/degree of nerve cord termination. The following differences were observed: 1) genitalia and intestinal appearance and bladder bulge in 1/15 (6.7%), 2) the presence of omphalocele in 2/15 (13.3%). From the initial radiological interpretation, 4/17 (23.5%) ultrasound and 2/19 (10.5%) MRI were misinterpreted as OEIS spectrum when the postnatal diagnosis was *Bladder Exstrophy*. Errors in diagnosis are caused by the bladder protruding beyond the plane of the abdominal wall with an intestinal loop in the posterior, imitating an omphalocele. In all cases, *Bladder Exstrophy* is a defect of the abdominal wall located in the inferior umbilical cord insertion on the abdominal wall.(Weiss et al., 2020)(Weiss et al., 2020)

Evangelidis et al. in 2004 reported a case of a 25-year-old G2P1 woman 33 weeks pregnant with a bladder that could not be visualized, bilateral identifiable kidneys, and amniotic fluid index within normal limits, indicating normal kidney function. Differential diagnosis includes *Bladder Exstrophy*, agenesis bladder, bilateral utter ectopic, urachus patent, and *non-visualization of the bladder*. The patient is planned to have a cesarean delivery, and the baby is born with *Bladder Exstrophy*. Routine prenatal ultrasound allows early diagnosis of major genitourinary malformations.(Evangelidis et al., 2004)

In another case, a report by Lee and Shim, a 28-year-old primigravida woman with a gestational age of 25 weeks was suspected of having a mass in the abdomen. Obstetric history and laboratory examination within normal limits. Ultrasound examination reveals a single fetus alive in a vertex presentation with biometrics consistent with gestational age. The homogeneous echogenicity mass measures approximately 2 × 2 × 4 cm and has an irregular, lobed surface protruding anteriorly at the bottom of the fetal abdominal wall (Figure 2). The bladder filled with normal fluid is not visible. The volume of amniotic fluid (AFI 13) is within normal limits. Both kidneys are very normal, even though the position of the left kidney is rather low. Interestingly, linear hypoechogenic structures on both sides of the abdominal mass were found. Color Doppler examination clearly identifies this as an umbilical artery (Kurjak & Zalud, 2020). Therefore, we conclude that the mass's origin is the bladder and are confident that the correct prenatal diagnosis is bladder exstrophy.(Lee & Shim, 2003)

Prenatal diagnosis *Bladder Exstrophy* It should be made in the presence of the following features: (1) a dense mass in the lower anterior abdomen, (2) an invisible fetal bladder, (3) a normal volume of amniotic fluid, (4) a low umbilical cord insertion, (5) a normal fetal kidney, (6) an outstretched iliac bone, (7) an umbilical artery located next to a protruding mass protruding from the lower abdominal wall and (8) an external genital malformation.(Lee & Shim, 2003)

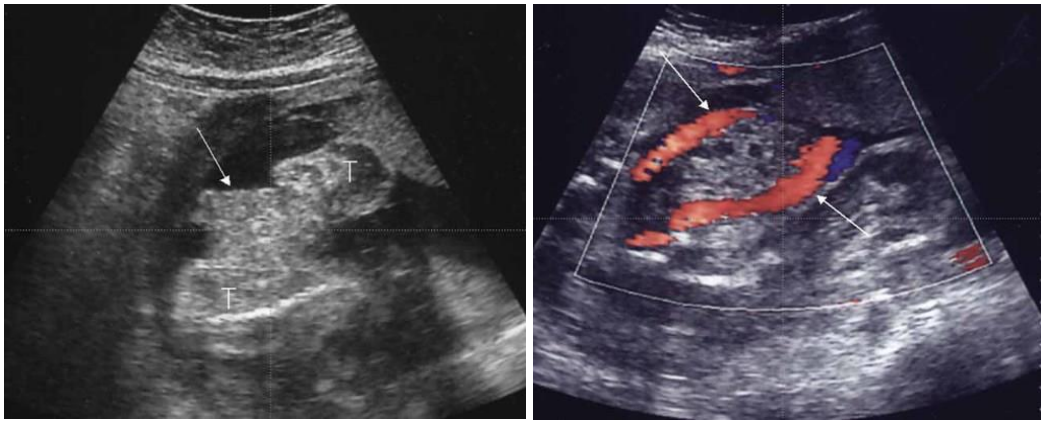
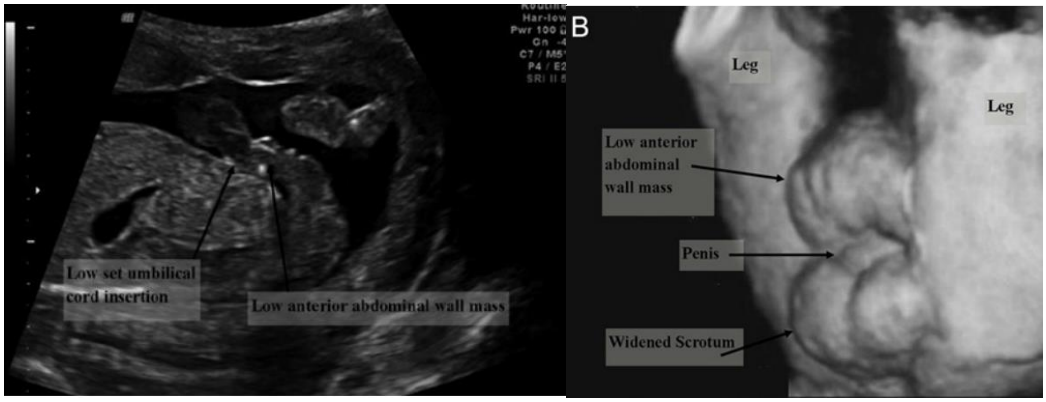
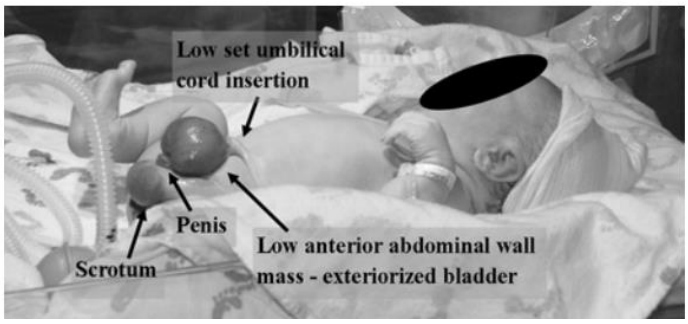


Figure 3. The coronal section of the anterior part of the lower abdomen of the fetus shows a dense mass (arrow) between the thighs (T)(Lee & Shim, 2003)
Source: processed data (2024)

In another case report by Wolniakowski et al., young women with G2P1 were given ultrasound examinations, including fetal biometrics consistent with gestational age, normal amniotic fluid, and normal fetal kidneys. The absence of the bladder, the inferior anterior abdominal wall mass of umbilical cord insertion, the insertion of the lower abdominal umbilical cord, the small penis, and the dilated scrotum (Figure 3a) were all noted, consistent with bladder exstrophy. 3D imaging (Figure 3b) aids diagnosis and improves patient understanding and conceptualization of fetal conditions. Findings at birth confirm the prenatal diagnosis, including inferior external bladder umbilicus. (Figure 3c).(Wolniakowski et al., 2014)



3a 3b



3c

Figure 4. *Bladder Exstrophy* (Wolniakowski et al., 2014)
Source: processed data (2024)

CONCLUSION

Bladder exstrophy is one of the rare and serious congenital disorders. Prenatal diagnosis of *Bladder Exstrophy* It can be difficult. It may be missed if the examiner does not allow enough time to fill the bladder and therefore considers that the absence of the bladder is physiological. In the absence of the bladder, other features may appear, such as a low umbilical cord and small external genitalia with an external mass of the lower abdomen and a dilation of the pubic ramus. Prenatal diagnosis allows the medical team to prepare for a multispecialty approach. It also gives parents time to prepare and adjust to expectations related to labor and the neonatal period, as well as potential complications and long-term prognosis.

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