Case report

EXTRAORDINARY BULGING MASS IN THE FOETUS - A CASE REPORT OF BLADDER EXSTROPHY

Authors:
Joanna Łosińska[1], Maria Respondek-Liberska[2,3]

1. Student of the Medical Univerisity of Lodz 2. Department of Prenatal Cardiology Polish Mother's Memorial Hospital Research Institute. Lodz, Poland. 3. Department of Diagnosis and Prevention of Fetal Malformations, Medical University, Lodz, Poland

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Abstract

INTRODUCTION: Prenatal diagnosis of bladder exstrophy is extremley rare and difficult.

BACKGROUND: Due to abnormal development of the cloacal membrane there is an incomplete closure of the lower abdominal wall, absence of the anterior wall of the bladder and external exposition of the posterior wall. The pubic bones are usually separated, the umbilical cord low inserted and there is abnormal external genitalia development.

CASE REPORT: At 21st week of gestation of 39-year-old multigravida multipara referred by a primary care obstetrician to high-specialised centre for a detailed ultrasound examination with a suspicion of bladder absence and inferior umbilical localisation. At 29 weeks of gestation presence of bulging mass of 2 cm, between the umbilical outlet and labia was detected. At 31 weeks of gestation previously detected structure among thighs had 3 cm diameter with lateral umbilical outlet. Major labia were prominent and minor labia were within normal limits. Between two umbilical arteries with an appropriate intraabdominal course there were no transsonic area corresponding to the urinary bladder. The newborn baby was born at term in a good condition, but with an exposed bladder of 4 cm in diameter. The urethral outlet was not visualised and the female genitals were abnormal. After a month the girl underwent primary bladder exstrophy closure. Although she suffers from recurring urinary tract infections, she is in a good general condition.

CONCLUSIONS: Due to prenatal diagnostics it was possible to detect and make an initial diagnosis of severe malformation. Early diagnosis allowed to prepare parents for a newborn with a defect and teach them how to take care of the baby.

Key words: *bladder exstrophy, prenatal diagnosis, ultrasound examination*

INTRODUCTION

Bladder exstrophy (BE) is a rare congenital complex anomaly with unspecified aetiology. Environmental mechanisms the same as genetics are suspected to be the causes. The incidence is recently reported to be 3 per 100 000 births[1]. Male to female ratio is mostly estimated to 2,5:1[2,3]. Due to abnormal development of the cloacal membrane there is an incomplete closure of the lower abdominal wall, absence of the anterior wall of the bladder and external exposition of the posterior wall. The pubic bones are usually separated, the umbilical cord low inserted and there is abnormal external genitalia development. In male patients BE is accompanied by epispadias. Female may suffer from vaginal outlet stricture, absence or doubling vagina the same as prolapse of reproductive organ. Although there are publications about post-natal management, surgical procedures and quality of life of those patients, prenatal diagnosis is scarcely described. Patients with BE face problems such as incontinence and sexual and reproductive difficulties. Early recognition enables to inform parents about therapeutical possibilities and estimated results of the treatment[4].

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CASE REPORT

A 39-year-old multigravida, multipara with unremarkable obstetric and family history, had first US exam at 14th week of gestation which was described as normal (Fig. 1). She was offered amniocenthesis for karyotype due to maternal age, but declined. At 21st week of gestation primary care obstetrician could not find a normal fluid-filled bladder and therefore the pregnant woman was referred for a detailed ultrasound examination to Wroclaw to The 2nd Department and Clinic of Gynaecology, Obstetrics and Neonatology. Inferior umbilical localisation and bladder absence was suspected in female fetus (Fig. 2).

Corresponding author: joanna.losinska14@wp.pl

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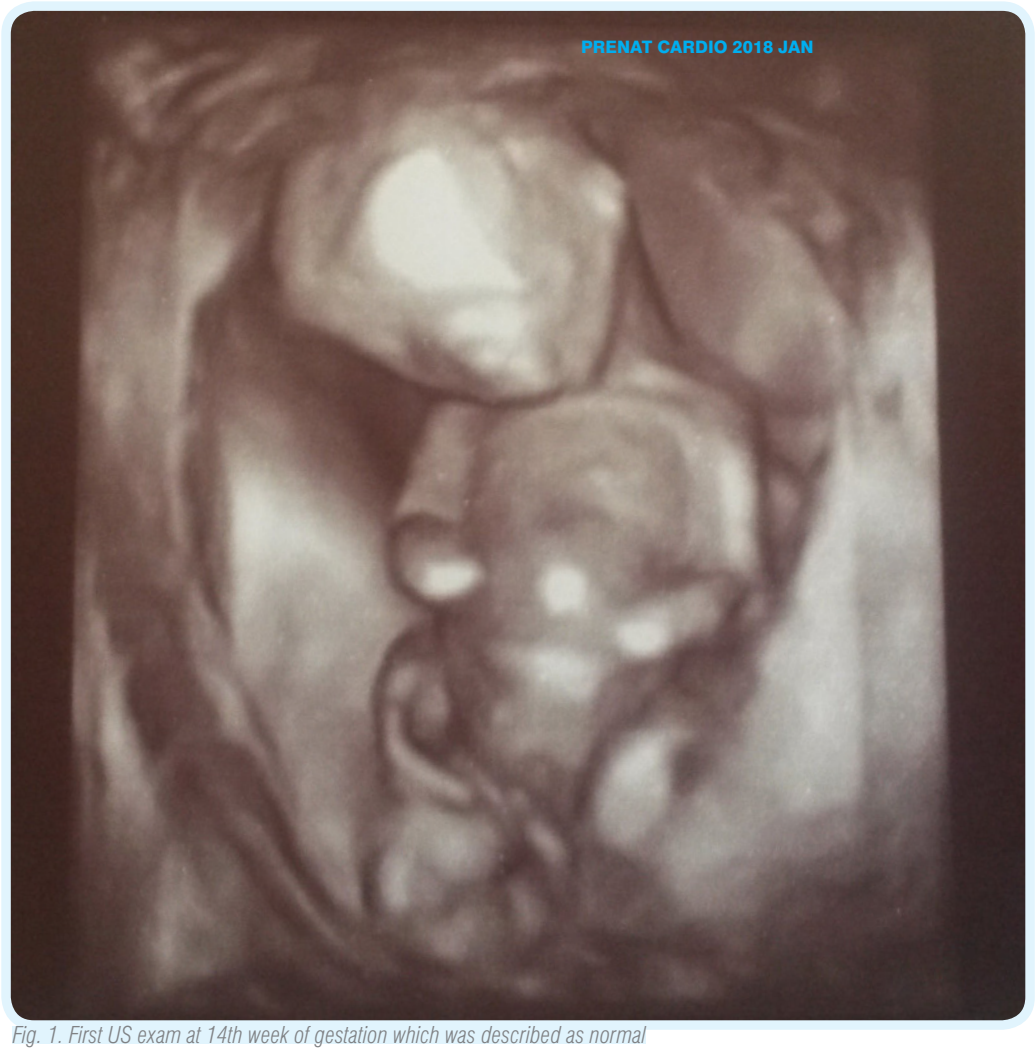

Fig. 1. First US exam at 14th week of gestation which was described as normal

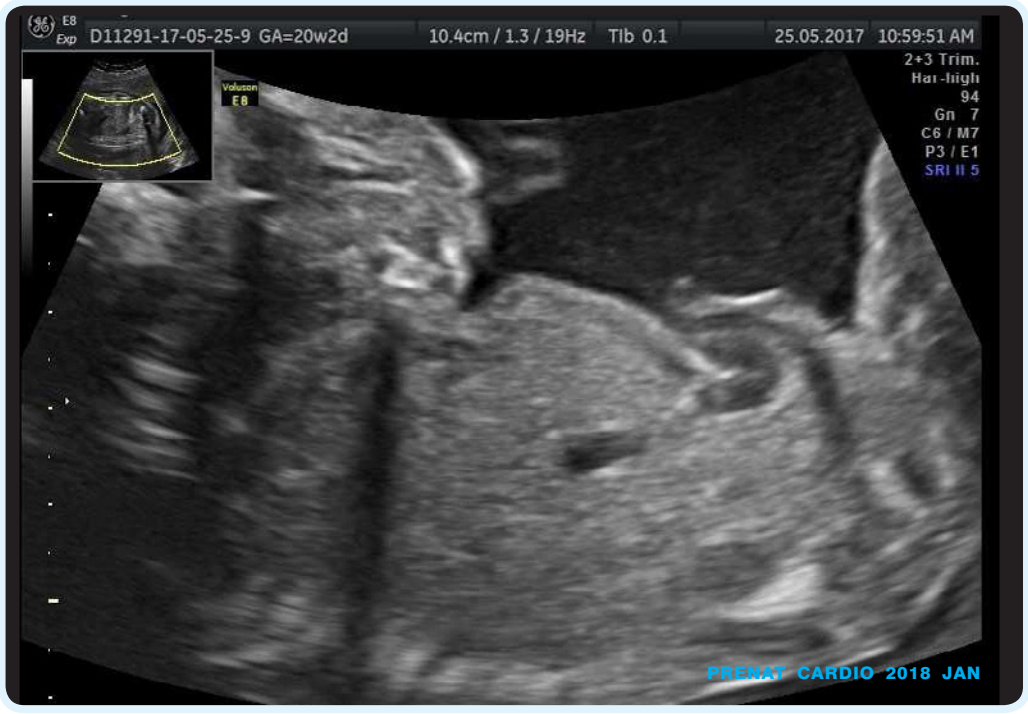

Fig. 2. At 21st week of gestation inferior umbilical localisation and bladder absence in female fetus.

At 29 weeks of gestation presence of bulging mass of 2 cm, between the umbilical outlet and labia was detected.

At 31 weeks of gestation gravida underwent sonographic and echocardiographic fetal examination in the referral centre in The Department of Prenatal Cardiology in Łodz. Previously detected structure among thighs had 3 cm diameter with lateral umbilical outlet. Major labia were prominent and minor labia were within normal limits (Fig. 3). Between two umbilical arteries with an appropriate intraabdominal course there were no transsonic area corresponding to the urinary bladder (Fig. 4).

There was normal fetal face (Fig. 5), normal heart anatomy and based on fetal echocardiography no functional abnormalities were present, normal both kidneys. The biometric parameters were consistent with gestational age. The newborn baby was born at 37 weeks of gestation by cesarean section with birth weight 3200g in a good condition, but with an exposed bladder of 4 cm in diameter. The urethral outlet was not visualised and the female genitals were abnormal. After 17 days of the hospital stay and maternal teaching how to take care of the newborn, she was discharged home and

referred to the Child's Memorial Health Institute in Warsaw. A month later the girl underwent primary bladder exstrophy closure. Although she suffers from recurring urinary tract infections, she is in a good general condition.

DISCUSSION

Prenatally recognised BE is reported infrequently and often ends up by termination of pregnancy[5-10]. Having BE increases 500 times the risk of this anomaly in the offspring[15]. In the Bladder Exstrophy in the fourth week of gestation mesenchymal cells seem to fail in migration between the ectoderm of the abdomen and the cloaca. Another theory says that this anomaly is secondary to pubic bones diastasis and the tension on the anterior abdominal wall leads to rupture[17,18]. Many of patients with BE are diagnosed postnataly[11,14]. The EUROSCAN study, a multicentre analysis of 709,030 births in 12 European countries, in which the total number of bladder exstrophy cases was 19, among which only 10 were diagnosed prenatally. Eight pregnancies were terminated, one resulted in a stillbirth, and 10 pregnancies ended with livebirths[14].

Goyal et al reported that in their study a quarter of 40 patients had antenatal diagnosis[11].

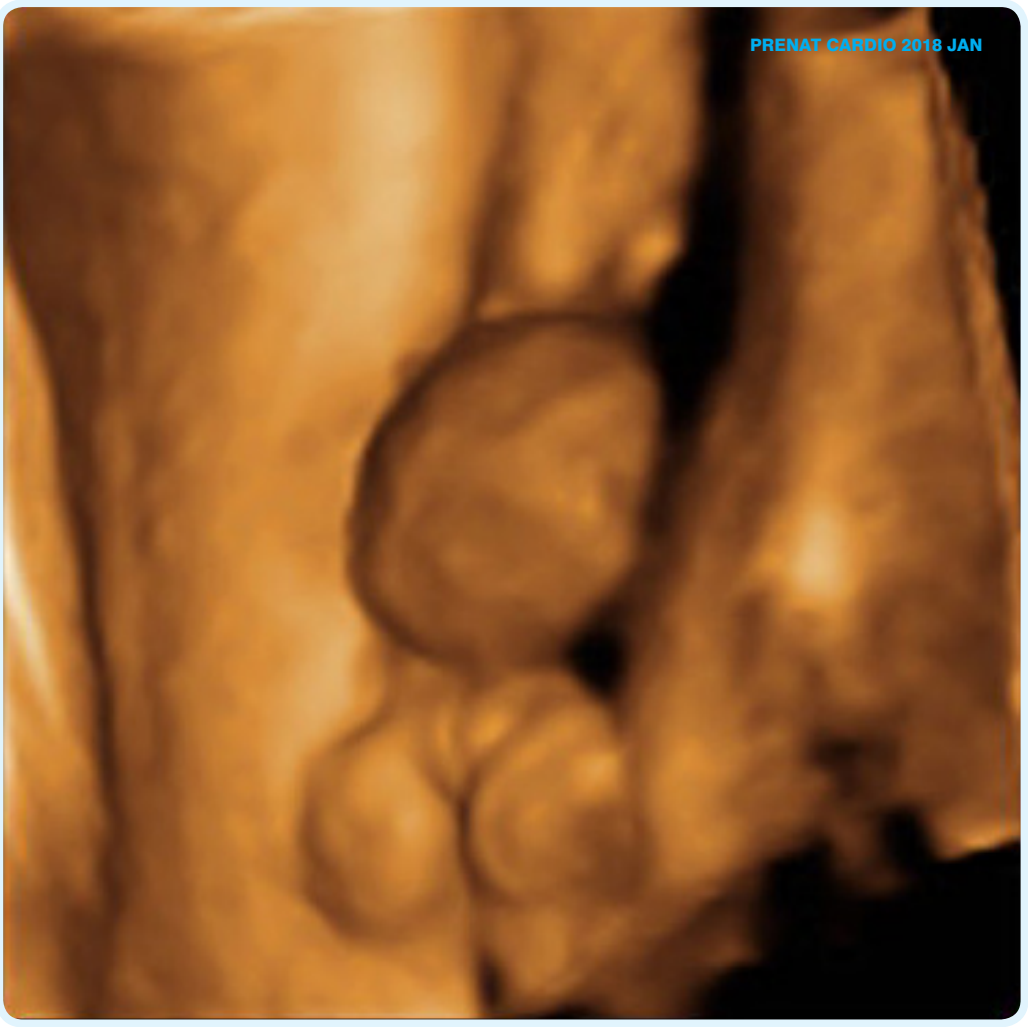

Fig. 3. Prominent major labia, normal minor labia.

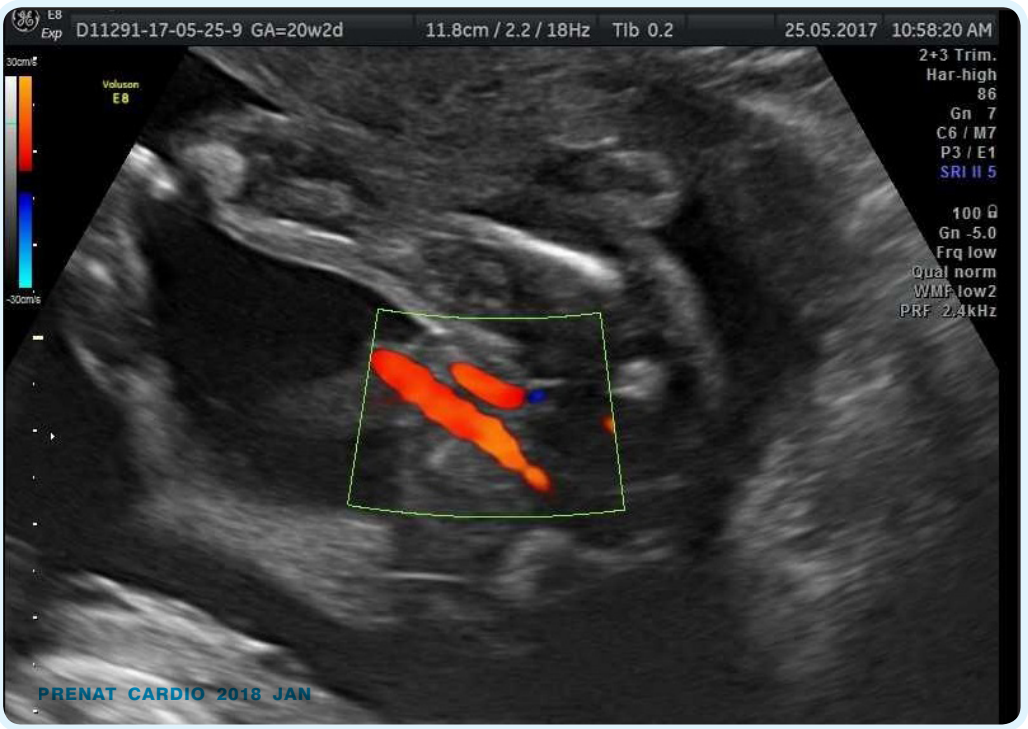

Fig. 4. No transsonic area corresponding to the urinary bladder arteries with an appropriate intraabdominal course

One of first disturbing sings is fetal urinary bladder absence. Normally it can be detected from[11-12] gestational weeks[13]. If unidentified till 15th hbd, can be considered as an abnormality. Although Goldstein et al described a case of bladder exstrophy in which the urachus resembled the normal bladder[10]. The differential diagnosis of bladder exstrophy includes omphalocele, gastroschisis but both with normally filled bladder and cloacal exstrophy in which concurrent bowel and genital abnormalities are seen[12]. An important point of prenatal recognition is visualisation by color Doppler ultrasound two umbilical arteries situated alongside the bulging mass in the lower abdominal wall. Normally those arteries originate from the external iliac arteries and run on both sides of the urinary bladder before entering the umbilical cord.

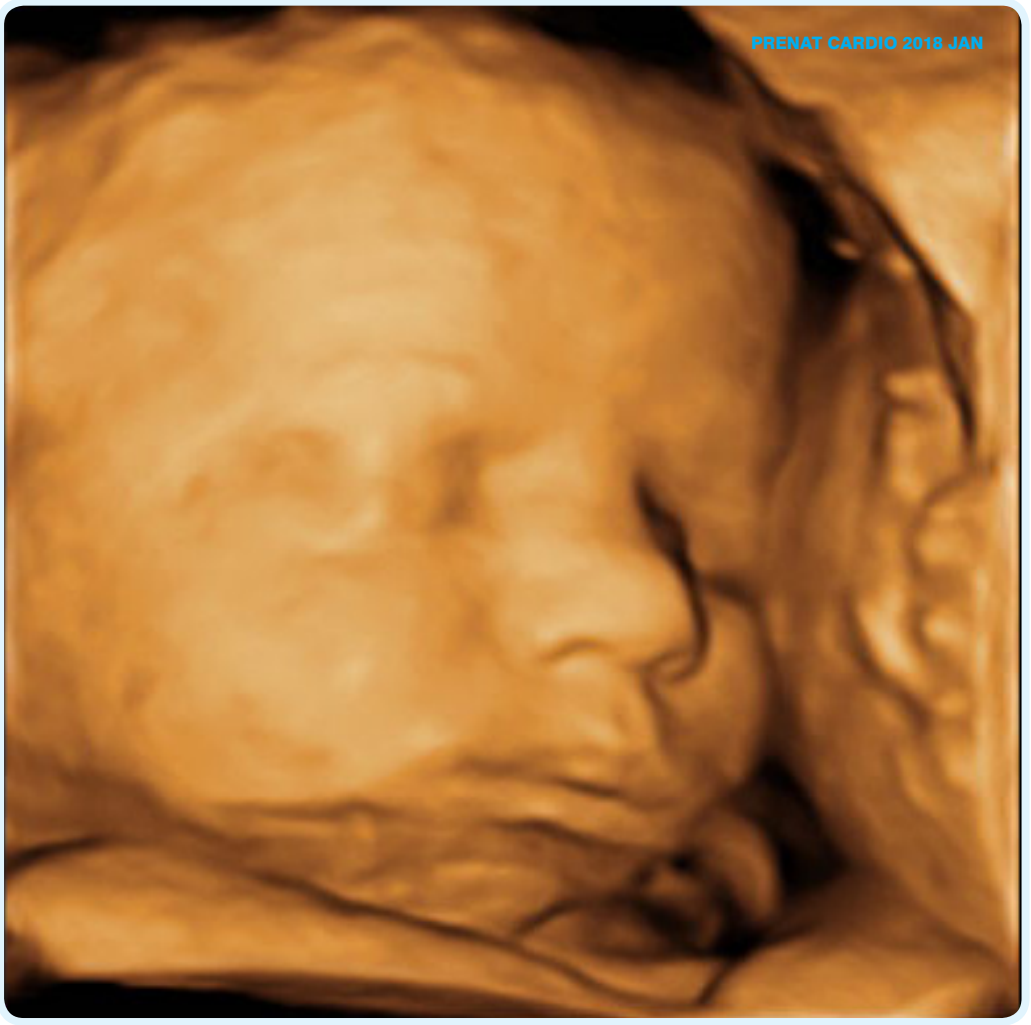

Fig. 5. Normal fetal face at 31 week of gestation.

Another disturbing observation in the reported case was low insertion of the umbilical cord. This feature may be considered as subjective and depended on the examiner but recent studies report that it also can be measured. Fishel-Bartal et al among 15 fetuses with nonvisualized bladder at age of 14-17 hbd correctly emerged 6 with BE by measuring umbilical cord-to-genital tubercle length. Malformation was diagnosed when the dimension was below the fifth percentile for gestational age[16].

We also would like to underline the value of prenatal detailed echocardiography evaluation in 3rd trimester. There was normal heart anatomy and no functional abnormalities which allowed to assume no necessity for premature delivery and good clinical outcome of the newborn baby without respiratory or heart problems[19,20].

Although the bladder exstrophy is not fatal, it has an enormous impact on patient's quality of life. Parents should be early informed about the malformation, possible management and it's prognosis. In the described case the family was fully aware of the problem and therefore the baby could be born, treated and operated on in high-specialised centres to minimalise complications and all of these were supported by appropriate parental care.

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J. Łosińska - first draft, literature search.

M. Respondek-Liberska - idea of the article, author of the 3D ultrasound pictures, final version

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