CASE REPORT

Diagnosis of Fetal Megacystis with Keyhole Appearance in Prenatal Ultrasound: A Case Report

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Abstract

A rare congenital condition known as fetal megacystis, characterized by an unusual enlargement of the fetal urinary bladder, exhibits a distinctive keyhole appearance. This anomaly was identified during a routine prenatal ultrasound of a male fetus at 21 weeks gestational age. The degree of fetal megacystis can vary, often exceeding the normal bladder measurement of 7–8 mm during the first trimester. The atypical configuration of the bladder and urethra, leading to a narrowed and elongated shape, imparts the keyhole appearance to the bladder. While the exact etiology remains uncertain, it is believed to be associated with various factors, including genetic disorders and urinary tract obstruction. This condition can profoundly affect the health and development of the affected fetus, potentially resulting in renal dysfunction, urinary tract infections, and organ damage. Other congenital anomalies, such as those involving the kidneys, lungs, or skeletal system, may also be linked to this condition. The management and treatment approach is contingent upon several factors, including the severity of the condition, the presence of associated anomalies, and the patient's gestational age. Close monitoring through regular ultrasound examinations may be recommended in milder cases to track the condition's progression. Interventions such as fetal urinary bladder catheter implantation or neurosurgical procedures may be viable options in more severe instances.

Keywords: Case report, Chromosomal abnormalities, Fetal megacystis, Fetal urinary bladder catheter placement, Posterior urethral valves, Prenatal diagnosis, Urinary tract obstruction.

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Introduction

Lower urinary tract obstruction is the primary contributor to fetal megacystis.[1] Detecting fetal megacystis during the initial stages of the first trimester of pregnancy typically portends a grim prognosis. In comparison, cases of posterior urethral valves (PUV) identified during the third trimester tend to exhibit a higher survival rate than those discovered during the second trimester.[1] Although ultrasound facilitates its identification, managing fetal megacystis remains arduous owing to its multifaceted etiology and uncertain progression. The likelihood of fetal megacystis occurrence is more pronounced in male fetuses than in female fetuses.[2] In instances where male fetuses exhibit fetal megacystis on ultrasound, the diagnosis commonly points to megacystis secondary to posterior urethral valve and prune belly syndrome.[2] Conversely, female fetuses often receive diagnoses of urethral atresia and megacystis-microcolon-intestinal hypoperistalsis syndrome.[2]

The measurement of bladder diameter emerges as a notable indicator for fetal megacystis. Antenatal ultrasounds for fetal megacystis indicate a bladder diameter exceeding 7 mm in the first trimester, 30 mm in the second trimester, and 60 mm in the third trimester. Additionally, antenatal assessments may reveal evidence of oligohydramnios and associated renal anomalies.[3] Among the most prevalent underlying diagnoses are PUV (57%), urethral atresia/stenosis (7%), prune belly syndrome (4%), megacystis-microcolon-intestinal hypoperistalsis syndrome (1%), and cloaca abnormalities (0.7%).[4] While ultrasonography can provide valuable insights, it is often insufficient to yield a definitive diagnosis but can strongly suggest a particular diagnosis.

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Case Presentation

This case report centers around a 24-year-old woman residing in a rural area. She was in her third pregnancy, with a history of two previous lower segment cesarean sections (LSCS). She sought antenatal care and visited the hospital's Outpatient Department (OPD). The fetal ultrasound examination revealed fetal megacystis, attributed to a posterior urethral valve. This condition was characterized by distinctive features such as an enlarged urinary bladder, a "keyhole feature," and a thickened bladder wall. Notably, the patient had no prior medical history of congenital anomalies, diabetes mellitus, tuberculosis, or hypertension.

The case is particularly noteworthy as it presents a diagnosis of fetal megacystis made via ultrasound at 22 weeks and one day of pregnancy. Specific indicators, including an elevated nuchal fold thickness and the presence of severe oligohydramnios, prompted this diagnosis. In this instance, the fetal megacystis was found to be secondary to the posterior urethral valve. As a result of these findings and the presence of suspected chromosomal abnormalities, the patient expressed her preference for pregnancy termination, which was subsequently performed through a hysterotomy.

Within the context of the second trimester, the anomalies scan report provided significant insights, including severe oligohydramnios characterized by a deficient liquor volume (0–1). Additionally, the report identified a sizable cystic lesion measuring 6.65 × 7.9 cm within the abdominal cavity. This cystic structure communicated with another cystic formation in the pelvis, presenting a keyhole appearance an indicative sign of fetal megacystis secondary to the posterior urethral valve. Furthermore, the report noted a markedly increased nuchal fold thickness measuring 12.5 mm.

Given the substantial megacystis and the suspicion of chromosomal abnormalities, the patient's decision to terminate the pregnancy was justified. This procedure was carried out through a hysterotomy.

Detailed fetal gestational parameters were provided during the ultrasound assessment, indicating specific measurements for biparietal diameter, head circumference, abdominal circumference, femur length, humerus length, ulna length, tibia length, and fibula length. The average gestational age was estimated to be 21 weeks and one day, with the fetal weight approximated at 530 grams, with an acceptable margin of error at ± 77 grams.

Observations during the ultrasound examination revealed a single, viable fetus in cephalic presentation. The fetal body movements and cardiac activity appeared within normal parameters, with a recorded fetal heart rate of 139 beats per minute. The placenta's location was anterior, displaying grade II maturity, and exhibited standard thickness and echo pattern, with no indications of previous placental concerns or hematoma. Notably, the liquor volume was severely reduced (0–1), resulting in restricted fetal movements due to insufficient amniotic fluid. The cervical length measured 42.7 mm, and both the internal OS and cervical canal were noted to be closed.

The impressions drawn from this comprehensive assessment indicated that fetal growth corresponded appropriately with the expected gestational age. While there was a mild increase in the cardiothoracic ratio, indicative of mild cardiomegaly, further evaluation of the heart, including the four-chamber view, outflow tracts, cardiac situs, and axis, revealed normal findings. Notably, both kidneys were deemed normal in size, location, and echogenicity, with no evidence of dysplasia or obstructive changes.

Regarding therapeutic interventions, amniocentesis was considered necessary to confirm any potential associated anomalies. However, given the patient's gravidity of 3 and her reluctance to undergo additional investigations, she decided to proceed with pregnancy termination through hysterotomy. The patient had previously undergone two LSCS (Figs 1 and 2).

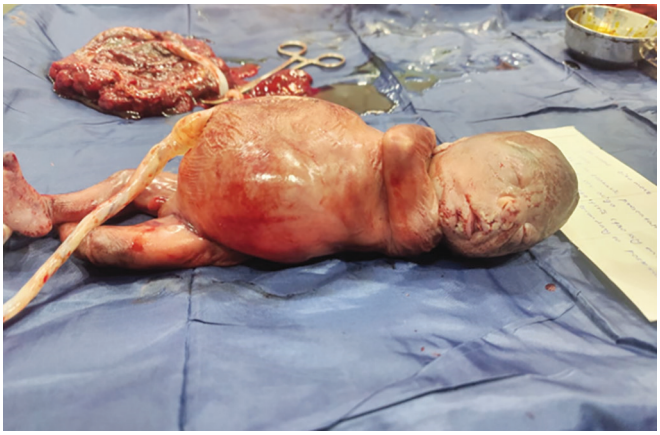

Fig. 1: Fetal appearance after labor induction. The fetus showed a grossly enlarged urinary bladder, suggesting fetal megacystis

Fig. 2: The placenta is anterior in location with grade II maturity and shows average thickness and echo pattern

Discussion

The causes of fetal megacystis can be classified into two categories: Nonobstructive and obstructive. Nonobstructive factors encompass conditions such as prune belly syndrome, megacystis-microcolon-intestinal-hypoperistalsis syndrome (MMIHS), chromosomal abnormalities, vesicoureteral regurgitation, and Neutrogena megacystis. On the other hand, obstructive causes include PUV or urethral malformations like stenosis, atresia, or persistent cloaca.[5]

This study illustrates that while lower urinary tract obstruction stands as the primary cause of fetal megacystis, it can also be accompanied by various genetic disorders, developmental defects, and chromosomal abnormalities, which result in an enlarged fetal bladder.[6] Unfortunately, this condition is associated with poor postnatal renal function and significant perinatal mortality.[7] Early fetal intervention in cases of severe first-trimester megacystis aims to minimize long-term renal damage, bladder dysfunction, intrauterine fetal death due to later anhydramnios, and neonatal death resulting from severe pulmonary hypoplasia.[8]

Fetal megacystis can have unidentified causes beyond typical fluctuations, and the outcomes differ depending on the underlying reasons. In cases without other complications, obstructive megacystis is treatable. Prior research has explored treatment options such as vesicoamniotic shunting, valve resection, and urinary stenting.[9] While vesicocentesis and vesicoamniotic shunts have been employed to manage severe megacystis in the first

trimester, their effectiveness in improving perinatal survival rates remains uncertain.[8]

In some instances, the pregnancy can be preserved, underscoring the importance of careful investigation into the specific causes of fetal megacystis. In the case discussed here, the fetus was at 21 weeks gestational age, and the primary diagnostic feature was the presence of fetal megacystis, evident through the prominently enlarged urinary bladder observed on ultrasound. The fetus displayed malformations, with the bladder exhibiting the keyhole sign and a thicker bladder wall, which are ultrasonographic markers indicative of posterior urethral valve-related fetal megacystis. Additionally, the fetus was male, and there was a notable reduction in amniotic fluid levels (oligohydramnios). This case was categorized as obstructive fetal megacystis due to posterior urethral valve obstruction and the keyhole appearance.[5] The presence of various structural anomalies in this case raised significant concerns about potential chromosomal abnormalities.

Moreover, increased nuchal fold thickness has been linked to chromosomal abnormalities.[10] In this particular case, nuchal fold thickness was significantly elevated, necessitating further investigation through amniocentesis to confirm the presence of associated congenital anomalies. However, the patient's obstetric history revealed that she was Gravida 3 Para 2 Live-2 (indicating three pregnancies, two deliveries, and two live births). Consequently, the patient was not prepared for further research, and a hysterotomy, a procedure involving the intact removal of the fetus from the uterus, was performed to terminate the pregnancy. Consequently, no additional investigations were pursued.

Conclusion

In conclusion, the management of fetal megacystis with a keyhole appearance, a rare and complex condition, necessitates the collaboration of maternal-fetal medicine specialists, pediatric urologists, and genetic counselors. Providing optimal care for affected fetuses and supporting their families throughout the journey requires precise diagnosis, comprehensive evaluation, and appropriate treatment. Ultrasonography is a vital tool in the diagnosis of fetal megacystis; however, it should not be the sole basis for a definitive diagnosis. Amniocentesis plays a crucial role in identifying chromosomal abnormalities, further enhancing diagnostic accuracy. To gain a deeper understanding of the underlying causes and to develop more advanced diagnostic and therapeutic strategies for this challenging condition, ongoing research, and collaborative efforts are imperative. Ensuring that affected fetuses receive the best possible care and that their families are well-supported remains paramount in the management of fetal megacystis with a keyhole appearance.

References

1. Bornes M, Spaggiari E, Schmitz T, et al. Outcome and etiologies of fetal megacystis according to the gestational age at diagnosis. Prenat Diagn 2013;33(12):1162–1166. DOI: 10.1002/pd.4215.
2. Osborne NG, Bonilla-Musoles F, Machado LE, et al. Fetal megacystis: Differential diagnosis. J Ultrasound Med 2011;30(6):833–841. DOI: 10.7863/jum.2011.30.6.833.
3. Weerakkody Y, Gupta S, Yap J, et al. Fetal megacystis. Reference article, Radiopaedia.org. DOI: 10.53347/rID-13417.
4. Taghavi K, Sharpe C, Stringer MD. Fetal megacystis: A systematic review. J Pediatr Urol 2017;13(1):7–15. DOI: 10.1016/j.jpurol.2016.09.003.
5. ISUOG. Fetal Megacystisis. Available from: https://www.isuog.org/clinical-resources/patient-information-series/patient-information-pregnancy-conditions/renal-system/fetal-megacystisis.html.
6. Fontanella F, Maggio L, Verheij JBGM, et al. Fetal megacystis: A lot more than LUTO. Ultrasound Obstet Gynecol 2019;53(6):779–787. DOI: 10.1002/uog.19182.
7. Fontanella F, Duin L, Adama van Scheltema PN, et al. Fetal megacystis: Prediction of spontaneous resolution and outcome. Ultrasound Obstet Gynecol 2017;50(4):458–463. DOI: 10.1002/uog.17422.
8. Ruano R, Yoshisaki CT, Salustiano EMA, et al. Early fetal cystoscopy for first-trimester severe megacystis. Ultrasound in Obstetrics & Gynecology. Wiley Online Library. DOI: 10.1002/uog.8963.
9. She F, Dong S, Yuan B, et al. Diagnosis of fetal megacystis with chromosomal abnormality by 2D prenatal ultrasound: A case report. Medicine (Baltimore) 2017;96(46):e8589. DOI: 10.1097/MD.0000000000008589.
10. Nicolaides KH, Wegrzyn P. Sonographic features of chromosomal defects at 11(+0) to 13(+6) weeks of gestation. Ginekol Pol 2005;76(6):423–430. PMID: 16149258.