Clinical Case Report

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# Diagnosis of fetal megacystis with chromosomal abnormality by 2D prenatal ultrasound

## A case report

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**Abstract**

**Rationale:** The pathogenesis of fetal megacystis is divided into obstructive and nonobstructive. Megacystis combined with chromosomal abnormalities is rare and most of the cases are nonobstructive.

**Patient concerns:** The fetus showed posterior urethral obstructive megacystis with features of bladder enlargement, "keyhole" feature, and thick bladder wall.

**Diagnoses:** Here, we present a case of fetal megacystis diagnosed by ultrasound at pregnancy week 15+2 and with multisystem abnormalities.

**Outcomes:** Moreover, the fetus showed edema, umbilical cord cyst, cardiac dysplasia, hook-shaped hand, and strephenopodia. These abnormalities strongly suggested chromosomal abnormalities. The fetus was diagnosed with trisomy 18 by amniocentesis. Posterior urethral obstructive megacystis was confirmed by pathology.

**Lessons:** In conclusion, this case suggests that in the presence of fetal megacystis and multisystem abnormalities, causes should be investigated and the possibility of chromosomal abnormalities should be considered in the presence of multisystem developmental abnormalities.

**Abbreviations:** hcg = human chorionic gonadotropin, MMIHS = megacystis-microcolon-intestinal-hypoperistalsis syndrome, NT = nuchal translucency, PUV = posterior urethral valves.

**Keywords:** chromosomal abnormality, fetal megacystis, posterior urethral valves, prenatal diagnosis, prune belly syndrome, vesicoamniotic shunting

## 1. Introduction

Fetal megacystis is an ultrasound sign with a detection rate of about 0.06%,[1] and with a male-to-female ratio of 8:1.[2] The ultrasound of normal fetal bladder usually demonstrates round or oval echo-free area in the fetal pelvic region. The fetal longitudinal diameter of the bladder during in the first trimester (11 to 13+6 weeks) is generally <6 mm.[3] If the diameter is ≥7 mm or if the bladder in the fetal pelvic region is large without emptying during a continuous observation of 45 minutes, then a diagnosis of megacystis may be given.[4] A variety of different causes can lead to megacystis, and these distinct causes result in different damage and prognosis of the fetal renal function. The most common etiology of megacystis includes posterior urethral valves (PUV) (57%), urethral atresia/stenosis (7%), and chromosomal abnormalities (15%).[2]

This case report evaluated a fetus of 15+2 weeks of pregnancy with megacystis and multisystem abnormalities. Taken together, these abnormalities were highly suggestive of chromosomal abnormalities, which was confirmed by amniocentesis. Moreover, megacystis combined with chromosomal abnormalities is generally of the nonobstructive type, while the present case showed megacystis and posterior urethral obstruction combined with chromosomal abnormalities, which is very rare.

## 2. Case presentation

This is a case report of a 27-year-old Chinese Han woman, gravida 2 para 0. Labor was induced in 2013 by administering C-class drugs without knowing the state of pregnancy. The patient had no adverse family history. At week 14+5, the patient received routine examination with abdominal 2 dimensional ultrasound at the Baodi District Beach Hospital of Tianjin. Ultrasound indicated megacystis and echo enhancement of both kidneys (but no fluid shown). Due to strong demands for fertility, the patient came to our hospital for re-examination at week 15+2. The ultrasound characteristics at week 15+2 are shown in Table 1.

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*FS and SD contributed equally to this work.*

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**Table 1**

**Ultrasound characteristics at week 15+2.**

| |
|---|
| Biparietal diameter: 2.8 cm |
| Head circumference: 11.1 cm |
| Abdominal circumference: 16.1 cm |
| Humerus length: 1.7 cm |
| Femur length: 1.5 cm |
| Abnormalities on ultrasound: |
| amniotic fluid of 3.0 cm |
| fissure with a width of 0.4 cm on the upper lip |
| defect of 1.5 mm in the ventricular septum |
| upper limb on one side was hooked |
| edematous skin |
| the thickest posterior cervical skin was 0.7 cm |
| the thickest abdominal skin was 0.8 cm |
| cystic mass of 1.1 cm at the site of the umbilical cord and entering into the abdominal wall |
| both renal pelvis were without significant expansion, but echo was enhanced in both kidneys |
| thickness of the bladder wall was 3.2 mm |
| cystic mass in the lower abdomen, $4.\times4.0\times3.1$ cm |
| "keyhole" sign |
| wrapped umbilical artery on both sides |
| fetal bilateral plantar and tibiofibula were in the same plane |
| distance from the lower border of placenta into the inside opening of the cervix was 2.6 cm. |
| The ultrasound suggested: |
| mid-term pregnancy at 14+3 wk; |
| multiple-system abnormalities (megacystis, cyst of umbilical cord, cleft lip, defect of ventricular septum, fetal edema, and abnormal development of limb); |
| considering the multiple-system abnormalities, chromosomal abnormalities could not be excluded. |

Considering the abnormal multiple-system development of the fetus, chromosomal abnormalities could not be excluded. The patient underwent puncture and biopsy of the amniotic cavity for karyotype analysis, which was 47XY, +18. Therefore, labor was induced. Anamnesis and examinations revealed regular menstruations; urine human chorionic gonadotropin (hcg) was positive (home test) at 28 days of pregnancy; ultrasound examination showed early intrauterine pregnancy of 50+ days; the actual week of pregnancy was 1 week less than the speculated week of pregnancy; early pregnancy reaction was not strong; the patient was without any history of tocolysis, radiation exposure, and vaginal bleeding; herpes virus, rubella virus, cytomegalovirus, and toxoplasma during pregnancy were negative. Down screening and amniocentesis were not done. At 3 days after admission (16+5 weeks), labor was induced. The fetus was male, with cleft lip and palate, angular deformity of both upper limbs, exstrophy of abdominal wall (cyst ruptured during labor), fetal edema, and developmental abnormalities of both lower limbs. Autopsy showed PUV (Figs. 1–4).

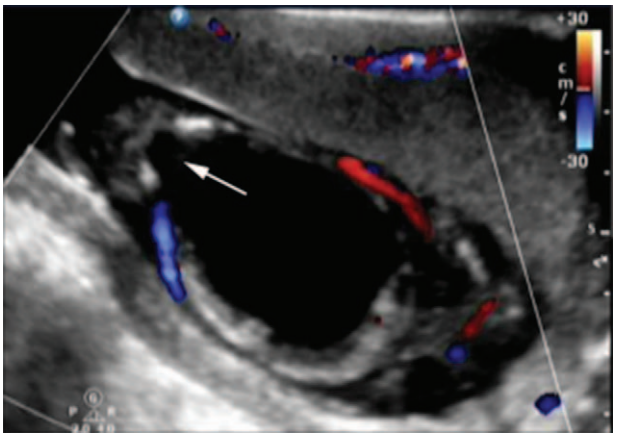


**Figure 1.** At week 15+2 of pregnancy, a large cystic mass in the pelvic region of the fetus was visible, with umbilical artery on both sides and the key hole sign (arrow).

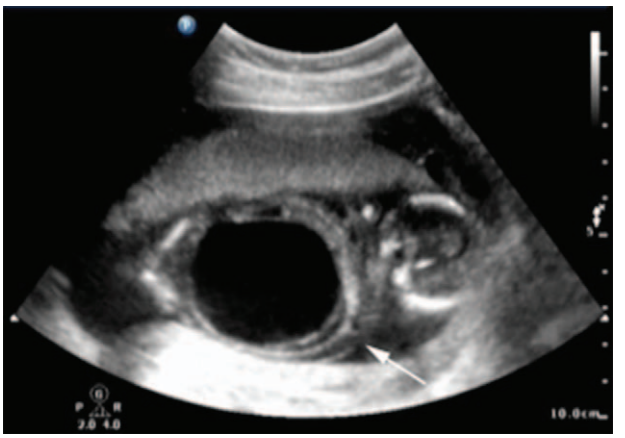

**Figure 2.** The fetal bladder wall was thickened (arrow).

## 3. Discussion

The causes of megacystis are divided into nonobstructive and obstructive. The nonobstructive causes include, among others, prune belly syndrome, megacystis-microcolon-intestinal-hypoperistalsis syndrome (MMIHS), vesicoureteral regurgitation, chromosomal abnormalities (mainly trisomy 13 and 18), primary giant urethra, and neurogenic megacystis. Obstructive fetal megacystis includes PUV, urethral atresia, and urethral stricture,[5] and there are some unknown causes of megacystis and a transient normal variation. Outcomes of distinct causes of megacystis are also different. Obstructive megacystis without other complications is treatable. Previous studies have already described the treatment methods, such as vesicoamniotic shunting,[6,7] valve resection,[8] and urinary stent.[9,10] Therefore,

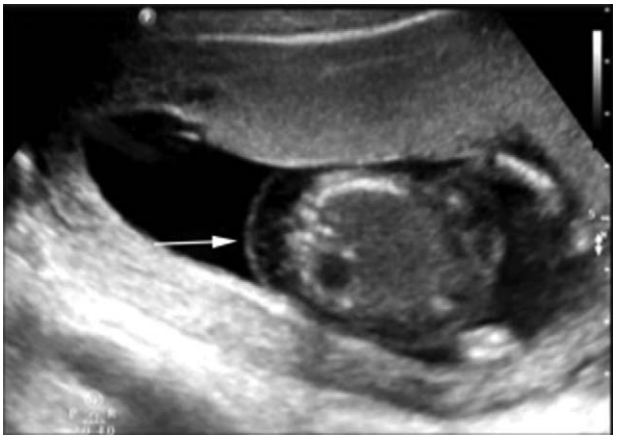

**Figure 3.** Abdominal wall edema of the fetus (arrow).

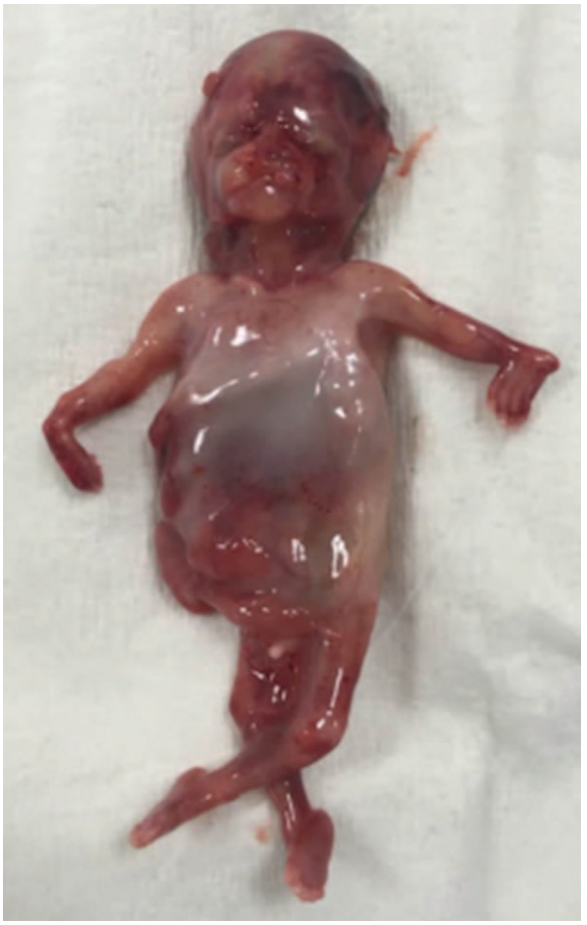

**Figure 4.** Fetal appearance after labor induction. The fetus showed cleft lip and palate, angular deformity of both upper limbs, exstrophy of the abdominal wall (cyst ruptured during labor), fetal edema, and developmental abnormalities of both lower limbs.

for women with strong demand of fertility, caution should be taken when investigating the specific causes of megacystis because the pregnancy can be saved. In this case, the fetus was at a young gestational age, and the most prominent feature for diagnosis was megacystis. Analysis showed that the fetus was basically in line with the ultrasound features of PUV caused by megacystis: enlarged bladder, thickened bladder wall, bladder with the keyhole sign,[11] the fetus was male, the amount of amniotic fluid was critical (but without oligohydramnios and hydronephrosis), and the injury to renal function due to megacystis was still in its early stage. Nevertheless, besides the megacystis, multisystem abnormalities were also found, including fetal edema, cyst of umbilical cord, heart dysplasia, hook hand, and strephenopodia, which are usually associated with chromosomal abnormalities.[12,13] Although many chromosomal abnormalities lead to fetal nonobstructive megacystis, the present case was highly suspicious of chromosomal abnormalities due to the multiple structural abnormalities. Therefore, it was necessary to conduct amniocentesis for further analysis. Moreover, it has been reported that chromosomal abnormalities are associated with nuchal translucency thickening.[14] If the effects of maternal age and nuchal translucency thickening are accounted for, the risk likelihood of trisomy 13 or 18 in the presence of megacystis increases by 6.7 times,[15] which is much higher than that of the general population. The woman did not undergo nuchal translucency examination at 11–13+6 weeks due to no adverse family history, which thereby missed the ideal opportunity for early detection of the abnormalities.

## 4. Conclusion

In summary, ultrasound examination is a very important method for the diagnosis of fetal megacystis. In the presence of megacystis and multiple fetal abnormalities, analysis for their causes should be performed, but the final diagnosis should not be reached based only on ultrasound examinations. Megacystis combined with multisystem development abnormalities should be considered suspicious of chromosomal abnormalities. The combined application of multiple examination methods could greatly improve the prenatal diagnosis of conditions causing megacystis.

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