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ORIGINAL ARTICLE

Fetal megacystis at 11–14 weeks of gestation: 3-year experience of a tertiary center

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ABSTRACT

Objective: We aimed to present a comprehensive overview of the underlying etiologies and structural anomalies associated with megacystis and to investigate the history from diagnosis in utero to postnatal outcome.

Material and Methods: Fetal megacystis was defined in the first trimester as a longitudinal bladder diameter ≥7 mm. For each case, data on and measurements of fetal urinary tract and associated structural anomalies were collected. All available postmortem examinations and postnatal investigations were reviewed to establish the final diagnosis.

Results: A total of 31 cases with fetal megacystis were included in this study. Megacystis was isolated in 24 cases and seven cases were associated with other abnormal ultrasound findings. Spontaneous resolution occurred before birth in 15 cases. Chromosomal abnormality was diagnosed in five cases, including two trisomy 21, one trisomy 18, one trisomy 13, and one Turner syndrome. Vesicoamniotic shunt was performed in three cases after the failure of vesicosentesis procedure.

Conclusion: The etiology of fetal megacystis is not known exactly. It is generally a temporary finding in the first trimester, and its continuation in the following weeks is observed with increased chromosomal anomaly and additional anomalies. In fetal megacystis, detailed ultrasonoography and chromosomal analysis should be performed for additional anomalies.

Keywords: Chromosomal abnormality, fetal megacystis, first trimester, prenatal diagnosis.

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INTRODUCTION

Fetal urine production begins at approximately 10 weeks' gestation, when the urinary bladder can be identified as an anechoic structure within the fetal pelvis, surrounded by the two umbilical arteries.[1] The finding a bulging urinary bladder, also known as fetal megacystis, is easily accomplished on ultrasound but is difficult to manage due to its diverse etiology and uncertain evolution. Fetal megacystis is most commonly defined as a longitudinal bladder diameter (LBD) ≥7 mm during first trimester, and in the second and third trimesters, it is generally as an enlarged bladder failing to empty during a period of at least 40 min as or a sagittal dimension in millimeter greater than gestational age in weeks +12 is often accepted. Beyond the first trimester, the prevalence of megacystis and its definition is still unclear.[2] Fetal megacystis that is an enlarged urinary bladder is variably defined and understood. The literature on fetal megacystis was systematically reviewed, focusing on prenatal diagnosis, associations, and outcomes. Fetal megacystis has an estimated first-trimester prevalence between 1:330 and 1:1670, with a male to female ratio of 8:1.[3] Early or late megacystis terminology was used according to gestational week before or after 18 weeks. Although the findings in the first trimester are usually temporary, their persistance or detection in the following weeks are associated with increased chromosomal and structural abnormalities.[1,2]

Megacystis can be associated with a thickened bladder wall defined as >3 mm. Oligohydramnios is present in approximately half of all cases. The main cause of fetal megacystis diagnosed in any trimester of pregnancy is bladder outlet obstruction, also known as lower urinary tract obstruction (LUTO).[4] In cases with severe early megacystis, parents often choose to terminate the pregnancy. Early megacystis with less severe cases and negative genetic diagnosis, a spontaneous resolution often occurs.[5] Although the main cause of fetal megacystis is LUTO, an enlarged fetal bladder can also be present as a concomitant finding of miscellaneous genetic syndromes, developmental disturbances, and chromosomal abnormalities. The spectrum of etiology and prognoses makes the counseling and management of this condition particularly challenging.[6] Given the low prevalence of fetal megacystis and the main focus on LUTO as etiology, the other causes of enlarged fetal bladder have been poorly investigated.[5] Vesicocentesis and vesicoamniotic shunt procedures can be used as treatment methods in the treatment of fetal megacystis, their effectiveness in improving perinatal survival rates has not been proven yet.[7]

The aim of this study is to provide an overview of the underlying etiologies and structural anomalies associated with fetal megacystis and to identify patterns of anomalies and ultrasound features related to specific complex anomalies beyond LUTO.

MATERIAL AND METHODS

This study was a national retrospective study carried out in the Perinatology Unit at İstanbul Kanuni Sultan Süleyman Research and Training Hospital between January 2018 and January 2021 years. Fetal megacystis was defined as LBD ≥7 mm during first trimester. The LBD was obtained from a sagittal view of the fetus, by measuring the distance from fetal bladder dome to bladder neck. Detailed anomaly scan was performed in all cases at the time of diagnosis in the first trimester, and karyotype analysis was offered for all cases. Parents were counseled about the prognosis of the condition and informed about the possibility of in-utero treatment. Vesicocentesis and vesicoamniotic shunt placement was only offered to chromosomally normal fetuses with isolated signs of LUTO and with concomitant oligohydramnios. Prenatal and postnatal data were collected in all cases. Nuchal translucency (NT) was measured at all cases at the first trimester examination and was considered increased if >95th percentile for the gestational age. NT measurement was collected retrospectively for fetuses referred later in pregnancy. Close ultrasound follow-up was offered to all cases who do not accept termination.

All available postmortem examinations and postnatal investigations were reviewed to establish a final diagnosis. LUTO was defined as a bladder outlet obstruction caused by urethral valves, urethral stenosis, or urethral atresia. Neonatal death was defined as death of a liveborn during the first 28 completed days postpartum. The megacystis was considered to be resolved if the bladder was observed to be empty following fetal micturition.

Vesicocentesis was performed first to all cases before vesicoamniotic shunt procedure. Vesicoamniotic shunt procedure was performed in cases developed megacystis again after the failure of vesicocentesis. Patients were followed weekly by ultrasound after shunt placement.

RESULTS

We identified 31 cases of fetal megacystis from the databases. The natural history, from diagnosis in utero to postnatal outcome, was reviewed. While 24 of the cases (77,4%) were isolated, the remaining 7 cases (22,6%) were associated with abnormal findings at ultrasonography such as increased NT in five cases, followed by single umbilical artery in two cases and cardiac defects in one case (more than one abnormal finding was observed in the same case).

While LBD measurement was below 12 mm in 13 cases (41.9%) in the study group, it was above 12 mm in the remaining 18 (58.0%) cases. Spontaneous resolution occurred in 15 cases (48.4%) before delivery.

Chromosomal defects were seen 5/31 cases with all LBDs below 12 mm. Chromosomal abnormalities were detected in five cases, including two trisomy 21, one trisomy 18, one trisomy 13, and one Turner syndrome. Vesicocentesis was performed before vesicoamniotic shunt in three cases. Vesicocentesis failed in three cases and vesicoamniotic shunt was applied to all of them who developed megacystis again. Table 1 shows characteristics and outcomes of the 31 cases of megacystis diagnosed prenatally.

Pregnancy results of 31 cases are as follows: 24 cases were born alive and two died in the neonatal period, four cases with severe megacystis or chromosomal anomaly were terminated, and one case resulted in fetal death in utero. A total of three cases accepted the shunt treatment (Table 2).

Table 1: Characteristics and outcomes of 31 cases of megacystis diagnosed prenatally

Cases	n
Isolated	24
Associated with other abnormal findings	7
Nuchal tranclucency	5 (>95p)
Longitudinal bladder diameter	13 (<12 mm)
	18 (>12 mm)
Antenatal intervention	3
Abnormal karyotype	5

Table 2: Characteristics of shunt cases

Cases	Intervention weeks	LBD (mm)	Outcome	Diagnosis after birth
1	16	17	No problem	37 week delivery
2	18	16	No problem	35 week delivery
3	21	31	Shunt dislocation	Fetal death

LBD: Longitudinal bladder diameter.

DISCUSSION

Fetal megacystis poses a challenge in terms of counseling and management because of its diverse etiology and evolution. Although the main cause of fetal megacystis is LUTO, an enlarged fetal bladder can also be present as a concomitant finding of miscellaneous syndromes, developmental disturbances, and chromosomal abnormalities. The main problem in the work-up of fetal megacystis remains its definition and the lack of a standardized antenatal management approach.[8]

The pathogenesis of fetal megacystis is divided into obstructive and non-obstructive. The authors described three possible evolutions with different outcomes: Chromosomal abnormalities, antenatal spontaneous resolution, and progression to obstructive uropathy. Outcomes of distinct causes of megacystis are also different.[9]

Megacystis with chromosomal abnormalities is rare and most of these cases are non-obstructive. Causes should be investigated in fetal megacystis and multisystem abnormalities, and the possibility of chromosomal abnormalities should be considered in multisystem developmental abnormalities. A variety of different causes can lead to megacystis, and these distinct causes result in different damage and prognosis of the fetal renal function. Due to the variable etiology, evolution, and prognosis, prenatal counseling in fetal megacystis is challenging.[10]

In the present study, we suggested karyotype analysis to all cases with fetal megacystis and although we found 16% percentage according to genetic anomalies with the most common chromosomal abnormality Trisomy 21, followed by trisomy 13 and 18. Liao et al.[11] demonstrated that there is a risk of about 25% chromosomal defect in fetal megacystis if LBD is 7–15 mm and 10% chromosomal defect risk with fetal megacystis higher than 15 mm. In fact, unlike our study, Syngelaki et al.[12] reported trisomy 18 as the most common chromosomal abnormality in first-trimester megacystis, followed by trisomy 21 and trisomy 13, whereas in the study by Liao et al.,[11] the most common chromosomal abnormality in fetuses with megacystis was trisomy 13, followed by trisomy 18, and less frequently trisomy 21.

In fetuses surviving the second half of pregnancy, LUTO commonly leads to hydronephrosis, renal dysplasia, and severe oligohydramnios, with a known poor diagnosis. LUTO was defined as a bladder outlet obstruction caused by urethral valves, urethral stenosis, or urethral atresia. The most common underlying diagnosis is posterior urethral valves (57%), followed by urethral atresia/stenosis (7%) and prune belly syndrome (4%). Karyotype anomalies are found in 15% including trisomy 18, 13, and 21 in the study by Fontanella et al.[13] This was similarly with our study.

In the present study, the termination rate of 13% was quite different from the study of Taghavi et al.[3] in which approximately 50% of fetuses with megacystis were terminated. Prognostic factors are oligohydramnios, gestational age at diagnosis, degree of bladder enlargement, renal hyperechogenicity, karyotype, and sex.[14] Oligohydramnios is the most important prognostic factors, including gestational age at diagnosis, degree of bladder enlargement, renal hyperechogenicity, karyotype, and gender.

In the present study, about 48% of the cases there was spontaneous resolution of the megacystis without any obvious adverse consequences on the development of the urinary system similarly to the study reported by Al-Hazmi et al.[15] Although there are similar rates in the literature, our high spontaneous resolution may be due to the low number of cases. However, our study showed that in some cases, the megacystis was associated with chromosomal defects and in others there was progressive obstructive uropathy.

Obstructive megacystis without other complications is treatable and previous studies have already described the treatment methods, such as vesicoamniotic shunting, valve resection, and urinary stent. Recently, thanks to technical improvements, early fetal therapy has, however, plausible as it appears that an early intervention is the only strategy potentially capable of preventing the occurrence of renal damage in the very few cases with truly isolated posterior urethral valves.[7]

Over the past few decades, fetal therapy before 18 weeks, in the form of fetal vesicoamniotic shunt and fetal cystoscopy, has been attempted with the aim of preventing early renal damage. Although vesicocentesis and vesicoamniotic shunts as treatment methods are used in the treatment of first trimester fetal megacystis cases, their effectiveness in improving perinatal survival rates has not been proven yet.[16] With early fetal cystoscopy, bladder dysfunction and renal damage can be prevented in fetuses with severe first trimester megacystic shunts caused by posterior urethral valve.[17]

In the present study, we selected suitable cases for early fetal therapy with normal NT, LBD >12 mm and without ultrasound evidence of any major abnormality. We performed vesicocentesis in all cases before shunt procedure and shunt procedure was per-

formed in all cases with recurrent megacystis. Recent studies have suggested that antenatal treatment improves perinatal survival.[18] The 33% dislocation rates in our study were nearly the same as those of Strizek et al.[19]

The authors generally suggested fetal karyotype analysis if LBD is between 7 and 15 mm. Spontaneous resolution will occur in 90% of these cases if the karyotype is normal. The prognosis is poor due to high incidence of LUTO in cases with LBD >15 mm. Since the study by Liao et al.,[11] there has been an increased interest in fetal therapy for LUTO.

When LUTO is suspected in the first trimester and megacystis is >12 mm, the prognosis is extremely poor and parents often opt for termination of pregnancy. For cases identified later in pregnancy, no definitive criteria for diagnosing LUTO and predicting the precise prognosis have yet been proposed.[18]

CONCLUSION

We presented the approach to fetal megacystis at 11 and 14 weeks of gestation in our clinic in the light of the current literature. The future studies are needed to optimize prenatal management and to prevent unnecessary fetal interventions.

Statement

Ethics Committee Approval: The İstanbul Kanuni Sultan Süleyman Health Training and Research Clinical Research Ethics Committee granted approval for this study (date: 10.03.0022, number: 2022.03.56).

Informed Consent: Written informed consent was obtained from patients who participated in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – GB, ÖÖ; Design – GB, ÖÖ; Supervision – GB, ÖÖ; Resource – GB, BD; Materials – GB, BD; Data Collection and/or Processing – GB, ÖÖ, BD; Analysis and/or Interpretation – GB, ÖÖ; Literature Search – GB, ÖÖ, BD; Writing – GB, ÖÖ; Critical Reviews – GB.

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