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Fetal megacystis across the gestational ages: The same sign for varying etiologies and outcomes

Lutfiye Uygur[1], Işıl Ayhan[1], Oya Demirci[1]

[1]Health Sciences University Zeynep Kamil Women and Children Diseases Hospital, Department of Obstetrics and Gynecology, Division of Perinatology İstanbul, Türkiye

Abstract

Objective: To compare the etiologies and perinatal outcomes of fetal megacystis according to their gestational age at diagnosis. The second aim is to detect the prognostic sonographic criteria in each gestational age group.

Methods: Fetal megacystis cases detected over eight years were retrospectively searched on hospital records. They were classified into three groups according to the gestational age at diagnosis: first, second, and third trimester of pregnancy. Pregnancy outcomes, postnatal prognosis, and etiologies were compared between the groups. The sonographic criteria that predicted the adverse outcomes were assessed for each trimester.

Results: A total of seventy-eight cases of fetal megacystis were included in the study. Thirty-two, forty, and six of the cases were diagnosed in the first, second, and third trimester respectively. 38.4% of the fetuses were born alive, and 19.2% of the fetuses survived beyond infancy. Gestational age at diagnosis, the presence of signs of renal impairment, and increased bladder diameter were associated with poor outcomes in the first trimester. Oligohydramnios, earlier gestational weeks at diagnosis, and an increased bladder length/biparietal diameter ratio were associated with poor outcomes in the second trimester. Third-trimester cases had better outcomes and were associated with varying pathologies.

Conclusion: The underlying etiologies of megacystis vary depending on gestational age at diagnosis. It may have a favorable prognosis when it presents in the later gestational ages and when abnormal amniotic fluid volume does not accompany. LBD: BPD ratio higher than 0.96 can be used as a poor prognostic factor in the second trimester of pregnancy.

Keywords: Megacystis, fetal malformations, urinary tract diseases, Lower Urinary Tract symptoms

Introduction

Fetal megacystis is the enlargement of the fetal bladder defined as a longitudinal diameter ≥ 7 mm in the first trimester.[1] Beyond the first trimester, the definition is not such clear. Although various methods exist to define megacystis in the second and third trimesters, the most accepted definition is 'an enlarged bladder that can not drain during an examination period of at least 45 minutes'.[2] The etiology and prognosis may differ according to the gestational age at which megacystis emerge and according to the additional ultrasonographic findings. The variability of underlying etiologies and prognosis is a challenge in prenatal counseling and management. This study aims to investigate the differences between the megacystis cases of three gestational age groups by comparing the ultrasound findings, underlying etiologies, perinatal and postnatal outcomes. The possible prognostic criteria will also be assessed for each trimester separately to contribute to the prenatal counseling of the families.

Methods

This retrospective cohort study was approved by the local ethics committee of an affiliated university hospital (24.11.2021, No: 176). It was conducted following the ethical standards described in an appropriate version of the 1975 Declaration of Helsinki, as revised in 2000. Informed consent stating that the data can be used for scientific purposes has been routinely provided from all

Correspondence: Lutfiye Uygur, Zeynep Kamil Women and Children Diseases Hospital, Department of Perinatology, İstanbul, Türkiye, **e-mail:** lutfiyeuygur@gmail.com, **Received:** January 29, 2024, **Accepted:** March 21, 2024

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ORCID ID: L Uygur 0000-0002-6325-1910; I Ayhan 0000-0002-8160-7853; O Demirci 0000-0001-5578-4437

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the patients who applied to the clinic. It was conducted in the Maternal Fetal Medicine (MFM) clinic of a single tertiary center. Fetal megacystis cases between January 2015 and December 2022 were searched on a computerized database The patients followed up in our maternal-fetal medicine center beginning from the first trimester and diagnosed with fetal megacystis in any of the ultrasound scans were included. The cases referred for fetal megacystis but not confirmed by MFM consultants were excluded. Monochorionic twin pregnancies, those who applied to our center after the first trimester of pregnancy, who did not comply with the follow ups, and those lacking hospital data were also excluded. The patients were grouped according to their gestational age at diagnosis: first trimester cases (10+0-13+6 gestational weeks), second trimester cases (14+0-27+6 gestational weeks), and third trimester cases (28+0- 41+6 gestational weeks). Ultrasound reports and images were reviewed.

In the first trimester, 'megacystis' was defined as '≥ 7mm for the longitudinal diameter of fetal bladder', and in the second and third trimesters, it was defined as 'an enlarged bladder which does not empty during the examination lasting 45 minutes and in the recurrent examination'. Gestational ages (GA) were confirmed with crown-row length (CRL) in the first trimester ultrasound. Oligohydramnios was defined as amniotic fluid index < 50 mm or deepest vertical pocket <20 mm. GA at diagnosis, longitudinal bladder diameter (LBD), CRL (for the first trimester), biparietal diameter (BPD) (for the second trimester), amniotic volume, urinary system (US) findings other than megacystis, renal cortical impairment findings (including hyperechogenic- dysplastic kidneys, cortical cysts, cortical thinning), extra urinary abnormalities, resolution of megacystis were recorded from the ultrasound reports and archives of the relevant images. Ultrasonographic signs of renal impairment in the first trimester were sought by transvaginal ultrasound examination per institutional protocol. Results of genetic studies were obtained from the electronic database of the 'clinical genetics' department. Termination of pregnancy (TOP) was approved by a multidisciplinary team based on genetic results, sonographic findings suggesting complex fetal malformations or sonographic findings suggesting renal failure such as dysplastic kidneys, oligohydramnios, anhydramnios with or without keyhole sign. Pregnancy outcomes, fetal and neonatal outcomes were sought from clinical records of the hospital computer database system. Defined etiologies and fetal and neonatal outcomes were compared between the gestational age groups. For each trimester, the patients were classified into two groups: long-term survival (survival beyond infancy) and poor outcome, including pregnancy terminations, fetal, neonatal, or infant demise. GA at diagnosis, LBD, US findings other than megacystis, abnormal extraurinary system findings, genetic anomalies, oligohydramnios, renal impairment signs, LBD/CRL (for the first trimester), and LBD/BPD (for the second trimester) were compared between these two groups to identify the prognostic criteria for each trimester.

Measures of association for categorical variables were analyzed with Chi-square and Fisher Exact test. Distribution patterns of continuous data were analyzed by the Shapiro-Wilk test. Skewed distributions of continuous variables in groups were compared using the Wilcoxon-Rank Sum test. Normally distributed continuous variables were compared by t-test. All analyses were performed using STATA software, version 17.0 Basic Edition (Copyright 1985-2021 StataCorp LLC). A p-value of <0.05 was considered statistically significant.

Results

A total of 84 cases were detected to have megacystis on routine ultrasound scans. Six of them were excluded due to a lack of hospital follow-up records. The mean maternal age was 28 years (± 6 y). The median gestational age at diagnosis was 15.2 weeks (range:10-36 w). Thirty-two, 40, and 6 cases were detected in the first, second, and third trimesters, respectively. Prenatal genetic studies could be employed for 56 fetuses. 19.6% of these fetuses had genetic abnormalities, including trisomy 18 (6 cases), trisomy 21 (2 cases), trisomy 13 (1 case), and del22.q.11.2 (1 case) (Table 1). A total of 32 cases (41%) had abnormal extraurinary system findings at diagnosis or follow-up ultrasound examinations. The most common associated abnormalities were congenital cardiac defects (32%), limb defects (26%), abdominal wall defects (19%), gastrointestinal defects (19%), nuchal thickness (16%), and single umbilical artery (16%). 41% of the cases presented oligohydramnios. In 44.8% of the cases, renal dysplasia findings were detected either in the first exam or in the follow-ups. 42.3% of the pregnancies were terminated on family request with the approval of the multidisciplinary committee based on ultrasonographic findings or genetic results. 19.2% of the cases died in utero. 38.4% of the fetuses were born alive, and 19.2% of the fetuses survived beyond the infancy period. Postnatal definitive diagnosis of the live-born cases differed from isolated uropathies to complex urorectal malformations (Table 1).

Table 1. Diagnosis of the underlying etiologies of fetal megacystis cases in the first, second, and third trimesters.

First trimester (11^{+0}-13^{+6} GW) n=32	Second Trimester (14^{+0}-27^{+6} GW) n=40	Third trimester (28^{+0}-41^{+6} GW) n=6
LUTO (n=13) (40.6%)	LUTO (n=25) (61.5%) VUR (n= 2)	LUTO (n=1) (PUV, partial bladder outlet obstruction) VUR (n=2) (VUR+ syringomyelia:1)
Chromosomal abnormalities (n= 6) (25%) -Tr21: (n=2) -Tr18: (n=3) -Deletion:(n=1) (del22q.11.2)	Chromosomal abnormalities (n=5) (12.8%) -Tr21: (n=1) -Tr18: (n=3) -Tr13: (n=1)	Chromosomal abnormalities (n=0)
Prune Belly Syndrome (n=1)	Caudal dysgenesis, anterior meningocele (n=1)	Unlabelled genetic syndrome (LUTO+ CDH) (n=1)
MMIHS (n=1)	MMIHS (n=1)	Detrusor hypoactivity (n=1)
Cloacal/urorectal malformation (n=2)	Cloacal/urorectal malformation (n=1)	Beckwith- Wiedemann (n=1)
Body stalk anomaly (n=1)	VACTERL association (n=1)	
Ehler Danlos Syndrome (n=1)	Miscellaneous syndrome (n=1)	
Miscellaneous syndrome (n=1)	Etiology not detected (n=2)	
Normal US (n=1)	Normal US (n=1)	
Etiology not detected (n=6)		

GW: gestational weeks, LUTO: lower urinary tract obstruction, Tr: trisomy, MMIHS: megacystis microcolon intestinal hypoperistaltism, US: urinary system, VUR: vesicoureteral reflux, CDH: congenital diaphragmatic hernia

*The families declined the autopsy or postmortem examination. Therefore, the diagnoses of LUTO were based on specific ultrasound findings like keyhole sign, early onset oligohydramnios, and renal dysplasia associated with megacystis in the terminated pregnancies or in case of intrauterine demise. The fetuses without either autopsy/postmortem examination or specific sonographic findings were classified as 'etiology not detected.'

When the etiologies of megacystis were compared between the groups, second-trimester cases were significantly more related to LUTO compared with the first-trimester patients. Chromosomal abnormalities and associated congenital syndromes did not differ between the groups (Table 2).

Table 2. The comparison of etiologies between the gestational ages

*Etiology/ associated abnormality	First trimester (11^{+0}-13^{+6} GW) n=32	Second Trimester (14^{+0}-27^{+6} GW) n= 40	Third trimester (28^{+0}-41^{+6} GW) n=6	P^1	P^2
Isolated uropathy - LUTO - VUR - Detrussor hypoactivity	40.6% (13/32) 13 (40.6%) 0 0	67.5% (27/40) 25 (62.5%) 2	66.6% (4/6) 1 (16.6%) 2 (33.2%) 1 (16.6%)	.06	.026
Chromosomal abnormalities	25% (6/24)	15.6% (5/32)	0 (0/6)	.35	.29
Associated congenital syndromes	24.1% (7/32)	12.5% (5/40)	33.3% (2/6)	.44	.46
Normal	3.1% (1/32)	2.5% (1/40)	0	.21	.13
Etiology not detected	15.6% (5/32)	5% (2/40)	0		

P^1 represents the difference between all three groups, and p^2 represents the difference between group 1 and group 2.
*The families declined the autopsy or postmortem examination. Therefore, the diagnoses of LUTO were based on specific ultrasound findings like keyhole sign, early onset oligohydramnios, and renal dysplasia associated with megacystis in the terminated pregnancies or in case of intrauterine demise. The fetuses without either autopsy/postmortem examination or specific sonographic findings were classified as 'etiology not detected.'

LUTO: lower urinary tract obstruction, VUR: vesicoureteral reflux, GW: gestational weeks

Regarding the outcomes, first-trimester cases were significantly related to intrauterine fetal demise compared with the other trimesters. Third-trimester cases were significantly associated with live birth, survival beyond infancy, and survival without renal insufficiency compared to the other GA groups (Table 3).

Table 3. Comparison of the outcomes of fetal megacystis cases detected in the first, second, and third trimesters

	First trimester (11^{+0}-13^{+6} GW) n=32	Second Trimester (14^{+0}-27^{+6} GW) n=40	Third trimester (28^{+0}-41^{+6} GW) n=6	P^1	P^2
TOP	37.5% (12)	52.5% (21)	0	NA	.20
IUFD	34.3% (11)	10.0% (4)	0	NA	.011
Live birth	28.1% (9)	37.5 % (15)	100% (6)	.004	.40
Survival beyond infancy	20.0% (5)	15.0% (6)	66.6% (4)	.009	.94
Survival without renal insufficiency	12.5% (4)	10.0% (4)	66.6% (4)	.002	.47

P^1 represents the difference between all three groups, and p^2 represents the difference between group 1 and group 2.

GW: gestational weeks; TOP: termination of pregnancy; IUFD: intrauterine fetal demise, N/A: not applicable

First Trimester Cases: This group had a median GA at diagnosis of 13 (10-13+6). The median LBD was 24 mm (8-65). Only 21.8 % of the cases presented oligohydramnios. One fetus with cloacal extrophy presented polyhydramnios on follow-ups. Twenty cases (62.5%) presented US findings other than megacystis, and 14 fetuses (43.7%) had abnormal extraurinary system findings on the initial or follow-up examinations. The most frequent associated extraurinary abnormalities were nuchal thickening and pes equinovarus. 43.7% of the fetuses presented renal impairment findings on abdominal or transvaginal USG. The outcomes are shown in Table 3. Diagnoses of the cases are shown in Table 1. None of the cases diagnosed with LUTO survived beyond the neonatal period in this group. When the ultrasonographic findings were compared between the group of fetuses with long-term survival and the ones with fetal, perinatal, or postnatal demise or underwent TOP, GA at the time of diagnosis, the presence of signs of renal impairment and increased LBD were associated with TOP or perinatal/postnatal death (Table 4).

Table 4. Prognostic criteria in the first trimester

Prognostic criteria	Total n, (%)	Survival beyond infancy	TOP/perinatal-postnatal demise	P value
Oligohydramnios	7 (100)	N/A	7 (100)	0.19
LBD (mm) (median, min-max)	24 (8-65)	10 (8-31)	25 (9-65)	0.048
LBD/CRL (median, min-max)	0.37(0.13-0.82)	0.23 (0.15-0.46)	0.37 (0.13-0.82)	0.08
US findings other than megacystis	20 (100)	2 (10)	18 (90)	0.25
Renal impairment signs	14 (100)	N/A	14 (100)	0.033
Abnormal EUF	14 (100)	3 (21.4)	11 (78.6)	0.42
Persistence of megacystis	26 (100)	2 (7.7)	24 (92.3)	0.012
GW at diagnosis (median, min-max)	13 (10-14)	13 (10-14)	13 (10-14)	0.37
Genetic abnormality	7 (100)	1 (14.2)	6 (85.8)	0.84

Data is represented as n (percentage) or median (range).
TOP: termination of pregnancy; N/A: not applicable; LBD: longest bladder diameter; CRL: crown-lump length; EUF: extraurinary findings, GW: gestational weeks, US: urinary system

Second Trimester Cases: The median GA at diagnosis was 17 (15-27) weeks. The median LBD was 39.5 mm (15-120). 77.5% of the cases had other US findings besides megacystis at the first appeal or follow-up examinations. 45% of the cases had renal cortical impairment findings, and 60% of the patients had oligohydramnios/ anhydramnios. Severe polyhydramnios emerged in one fetus during the follow-ups. 40% of the cases had abnormal extraurinary system findings on ultrasound. 5 (15.6%) of the cases who opted for genetic investigations were diagnosed with aneuploidies, including trisomy 18 (3 cases), trisomy 21 (1 case), and trisomy 13 (1 case). Megacystis resolved in three cases, one of which reoccurred due to obstruction of the vesicoamniotic shunt implemented. The second one

had a completely normal urinary tract in the follow-ups, and a healthy male baby was born at term. The third case was diagnosed with trisomy 18 and underwent TOP despite the resolution of megacystis. Perinatal outcomes of the fetuses diagnosed in the second trimester are shown in Table 3. When the prognostic criteria were assessed, oligohydramnios, earlier GA at diagnosis, and an increased LBD/BPD ratio were associated with perinatal/postnatal death or TOP (Table 5). The estimated cut-off of the LBD/BPD ratio for poor prognosis was 0.96 (Figure 1, Table 6).

Table 5. Prognostic criteria in the second trimester

Prognostic criteria	Total n, (%) median (min-max)	Survival beyond infancy (n=6)	TOP/perinatal- postnatal demise (n=34)	P value
Oligohydramnios	24 (100)	1 (4.1)	23 (95.9)	0.019
LBD (mm)	46.2 (15-120)	43.5 (17-50)	39 (15-120)	0.9
LBD/BPD	1.2 (0.26-3)	0.79 (0.51-1.34)	1.28 (0.26-3)	0.02
US findings other than megacystis	31 (100)	5 (16.2)	26 (83.8)	0.7
Renal impairment signs	18 (100)	2 (11.1)	16 (88.9)	0.53
Abnormal EUF	16 (100)	1 (6.3)	15 (93.7)	0.2
Persistence of megacystis	37 (100)	5 (13.5)	32 (86.5)	0.35
GA at diagnosis (weeks)		21.5 (16-26)	17 (14-27)	0.028
Genetic abnormality	5 (100)	N/A	5 (100)	0.3

Data is represented as n (percentage) or median (range).
TOP: termination of pregnancy; N/A: not applicable; LBD: longest bladder diameter; BPD: biparietal diameter; GA: gestational age, EUF: abnormal extraurinary findings; US: urinary system

Table 6. LBD/BPD cut-off value predicting poor outcome

Predictor	AUC	Cut-off value	Sn	Sp	PPV	NPV
LBD/BPD	0.79	0.96	%74.2	%80	%95.8	%33.3

LBD: longitudinal bladder diameter, BPD: biparietal diameter, AUC: area under curve

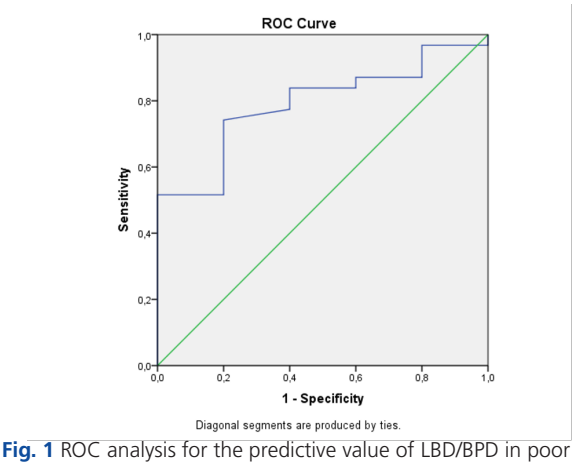

Fig. 1 ROC analysis for the predictive value of LBD/BPD in poor outcomes.

Third Trimester Cases: Eight cases were detected in the third trimester and two were excluded. The mean gestational age at diagnosis was 33.2 2.6. The median LBD was 80 mm (34-90). One of the cases had oligohydramnios, and two had polyhydramnios on ultrasound. Two cases had a thin renal cortex due to hydroureteronephrosis, and one had dysplastic kidneys with cortical cysts. All of the fetuses were live-born. Four of them survived beyond infancy. One neonate had CDH in addition to PUV and died in the first week of life. The other case was scheduled for renal transplantation. However, he died after corrective surgery for the transposition of great arteries, which had been missed in prenatal examinations. Ultrasound findings, postnatal diagnoses, and prognosis are shown in Table 7.

Table 7. Prenatal ultrasound findings, postnatal diagnoses, and prognosis of the third-trimester cases

Case no	Additional US findings (on USG)	Abnormal extraurinary findings (on USG)	Postnatal diagnosis	Postnatal additional anomalies	Prognosis
1	Hydroureteronephrosis	None	VUR	Syringomyelia	Operated for VUR, no renal impairment
2	None	Polyhydramnios macrosomia	BWS	None	No renal impairment
3	Hydroureteronephrosis	None	VUR	None	No renal impairment

4	Hydroureteronephrosis	Oligohydramnios	Incomplete PUV	TGA	Died in postnatal 6th month
5	Hydroureteronephrosis, Hyperechogenic, cystic kidneys	CDH, oligohydramnios	PUV+CDH	None	Mortality in the postnatal first week
6	None	Polyhydramnios	Detrussor hypoactivity syndrome	None	Postnatal intermittent catheterization, no renal impairment, normal urination after three months.

US: urinary system, USG: ultrasonography, VUR: vesicoureteral reflux, BWS: Beckwith Wiedemann Syndrome, PUV: posterior urethral valve, CDH: congenital diaphragmatic hernia, TGA: transposition of the great arteries,

Discussion

Megacystis is an ultrasound finding with various underlying etiologies with various prenatal and postnatal outcomes.[3] Our study shows that the most common etiology is LUTO in all GAs. Second-trimester cases were significantly more related to LUTO than first-trimester cases (Table 2). The third-trimester group also had similar isolated uropathy rates with the second-trimester group. However, VUR was the most profound etiology, and no cases of complete LUTO were detected in this group (Table 1, 2). This was anticipated as complete LUTO usually alerts the sonographer in earlier weeks with severe oligohydramnios. Both non-obstructive and obstructive uropathies can present megacystis with oligohydramnios in the second and third trimesters, depending on the degree of the pathology.[4]

Although the difference between the GA groups was not statistically significant, chromosomal abnormalities were more associated with the first trimester onset of megacystis. As the GA at diagnosis advances, chromosomal abnormalities occupy less part among underlying etiologies. In the study conducted by Bornes M. et al., the LUTO rate in the third-trimester megacystis cases was quite higher than in this study.[5] However, it was not stated in the methods whether the GA at diagnosis had been the week of the first appeal in their center or the first diagnosis had been made anywhere else before, for a particular pregnancy. Some cases of LUTO in the third-trimester group might have been diagnosed many weeks later than the first onset of sonographic findings. Only one third-trimester patient (the one with a diaphragmatic hernia with incomplete PUV) had missed her second trimester scan and had inadequate antenatal care in our study. She would probably had been diagnosed with LUTO in earlier gestational weeks with an earlier scan. Therefore, her GA group raises doubts. The remaining study group consisted of the pregnancies followed up in our center beginning from the first trimester. Thus, the GA groups could be established accurately in our study.

The rate of extraurinary system abnormalities and chromosomal abnormalities were quite high (43.7% and 25%, respectively) in the first trimester compared to the other studies in the literature.[6] Indeed, these two results depend on each other as a recent study reported that isolated megacystis without any extraurinary ultrasound findings was not associated with aneuploidies.[7] We detected renal cortical impairment findings in 43.7% of the first-trimester pregnancies, all of which had LBD> 15 mm. Previous studies did not report clear data about the renal cortical impairment signs in the first trimester. Lesieur et al. detected abnormal renal cortical findings in 14.7% of the megacystis cases in the initial ultrasound.[7] The rate was increased to 65.5% in the second examination. Our higher rate in the first ultrasound can be related to our adherence to the systematic transvaginal ultrasound examination in all of these cases. Despite the quite close rates of sonographic renal impairment signs between the first-trimester and second-trimester groups, the rate of oligohydramnios in the second-trimester group doubled that of the first trimester. This is obviously secondary to the evolution of amniotic fluid dynamics through the gestational weeks.[8] This fact also explains why oligohydramnios was detected as a poor prognostic factor in the second-trimester group but not in the first-trimester group. Another well-defined prognostic factor in the first trimester was LBD in the literature.[9] As it may also vary between gestational weeks, LBD: CRL rate was suggested as a valuable prognostic factor.[6] This variability is expected to be more obvious in the second trimester. Therefore, we investigated whether the LBD: BPD ratio could be used to predict prognosis. We found that LBD: BPD ratio was significantly increased in the poor prognosis group, and we detected a cut-off value of 0.96. Undoubtedly, the second-trimester cases present more self-explanatory signs representing the underlying pathology when compared with the first-trimester cases. However, as the performance of prenatal ultrasound in explaining the pathology and prognosis is still a matter of debate, and various sonographic clinical scoring models are being suggested[4], we suggest that LBD: BPD can be

implanted in these models and be employed for predicting the prognosis. This finding should be studied in future investigations with larger sample sizes.

Although statistically insignificant, the TOP rate was higher among the second-trimester group than in the first trimester. This can be explained by the fact that intrauterine fetal demise was significantly higher in the first trimester pregnancies. Obviously, some of the first-trimester cases experienced fetal death before the genetic results or before the sonographic criteria for the decision of TOP were completely established. The live birth rate increased with the GA. The difference was not statistically significant between the first and second-trimester groups; however, third-trimester cases were significantly related to live birth, survival beyond infancy, and survival without renal insufficiency. These results were in accordance with similar studies in the literature.[5,10,11] This reflects the fact that complete LUTO or complex malformations, which are expected to result in renal insufficiency, oligohydramnios, pulmonary hypoplasia, and other severe extraurinary system abnormalities present typical sonographic findings at earlier GAs. In accordance with the literature, both of the LUTO cases in the third trimester in our study group were incomplete PUV and did not present severe oligohydramnios.[12]

We had very few cases in the third trimester. Except for the one with inadequate follow-up, third-trimester cases had more subtle sonographic findings and, as a consequence, had more promising short-term outcomes. The only obstructive pathology was incomplete, and the infant died secondary to postoperative complications of congenital heart disease unrelated to renal insufficiency. Late-onset obstructive pathologies may also occur secondary to the masses in the fetal pelvis that enlarge by the gestational weeks. In that case, the prognosis depends on the original pathology that leads to megacystis, and renal functions are not expected to be severely impaired.[13] Therefore, we can inform the patients that isolated megacystis detected in the third trimester usually has better outcomes; however, it should be kept in mind that non-chromosomal genetic syndromes and neuromuscular dysfunctions may present in late gestational weeks.[3]

One of the weaknesses of our study is that we could not subclassify the obstructive pathologies. The families of the cases terminated, and fetal demises declined autopsy due to cultural and religious issues. Therefore, the pathology did not confirm the underlying etiologies, and the diagnoses were mostly based on the combination of the most specific sonographic findings like oligohydramnios, keyhole sign, and cystic and dysplastic kidneys in those cases. The second weakness of the study is that most of the genetic workup did not include chromosomal microarray and whole exome sequencing. On the other hand, almost all cases were followed up in the same tertiary center beginning from the first trimester, which assures the exact timing of the onset of the sonographic findings. Live-born cases were also all followed up in the same center.

Conclusion

It is already known that the first-trimester megacystis cases have a worse prognosis due to associated chromosomal and non-chromosomal anomalies, and amniotic fluid volume does not contribute to the prediction of prognosis. This study detected that renal impairment findings can be detected at a higher rate in the first trimester cases with transvaginal ultrasound to predict the prognosis earlier. This study also detected that an LBD: BPD ratio higher than 0.96 can be used as a poor prognostic factor in the second trimester of pregnancy. The gestational age at the onset of megacystis reflects the underlying etiology to some extent, and as a consequence, it impacts fetal and postnatal outcomes. As the gestational weeks at diagnosis advance, the worst scenarios, like chromosomal abnormalities and complete lower urinary tract obstruction, fall behind. The literature lacks megacystis cases that first present in the third trimester. This study showed that rare conditions like overgrowth syndromes or detrusor hypoactivity syndrome might also be the underlying pathology in the third trimester in addition to incomplete PUV and VUR.

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