Prenatal Diagnosis of Megacyst In Early Pregnancy: A Case Report

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A case with an incidental ultrasonographic finding of a megacysty in the 14th week of pregnancy, leading to an early diagnosis of Trisomy 18 is presented.
An 18-year-old primigravida attending to outpatient clinic for her routine antenatal check up at the 14th week of her pregnancy was observed to have a fetus with a 12 mm bladder. There were no other apperant major organ or system abnormalities. Under the guidance of current literature, the family was counseled by obstetrics and genetics departments, and a chorion villus sampling was adviced. The result of the CVS was reported as Trisomy 18. The family's wish of termination of the pregnancy was evaluated and the medical abortion with misoprostol induction was achieved.
When evaluating and screening an early pregnancy with ultrasound for fetal abnormalities, the size of the fetal bladder should be evaluated carefully and megacystic cases should be kept in mind with the possibility of chromosomal abnormalities.

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Key Words: Early pregnancy week, Megacyst, Prenatal diagnosis, Chromosomal abnormalities

There are wide series on the 2. and 3. trimester obstructive uropathies available in the literature.[1-3] On the contrary, there are limited data and literature on the megacyst findings at the late first trimester and early second trimester, and concomitance of other chromosomal and structural deformities.[4] A megacyst is defined as a fetal bladder which is 7 mm or more in longitudinal diameter between the 10th to 14th gestational weeks.[5] In a series of 24492 singleton pregnancies, 15 cases of megacyst has been diagnosed. Of those, 3 had chromosomal abnormalities, 7 cases have been resolved spontaneously by the 20th week of pregnancy, 4 cases have developed severe obstructive uropathies, and 1 case has been aborted.[5] We aimed to present a case of fetal megacyst detected in early pregnancy at our outpatient clinic under the current literature.

Case Report

An 18-year-old primigravida at the 14th week of her pregnancy, has been detected to have a fetus with a 12 mm bladder during routine antenatal ultrasound (Figure 1). Other fetal organs and systems did not present detectable major abnormalities (Figure 2). Along with the current literature, the family was counseled by genetics and obstetrics departments, and a chorion villus sampling was planned. The result was reported as Trisomy 18. Family's will of termination of the pregnancy was respected and the medical abortion of the pregnancy was achieved through misoprostol induction.

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Figure 1: Ultrasonographic view of a fetus with a 12 mm bladder

Figure 2: Other fetal organs and systems did not present detectable major abnormalities

Discussion

If a bladder diameter of 7–15 mm is detected between the 10th to 14th weeks of pregnancy, a chromosomal abnormality chance around 25% has been reported.[6] Trisomies 13 and 18 are prominent among those. If no chromosomal abnormality is detected, then there is a chance of 90% remission without any significant urological disorder.[6] A bladder diameter 15 mm or more has a chromosomal abnormality chance of 10%, but in those cases, the possibility of progressive obstructive uropathy later in the pregnancy is very high.[6]

Favre et al.[7] found trisomies 13,18 and 21 in 4 out of 5 cases with a bladder diameter between 9-15 mm, but reported no chromosomal abnormalities in 10 cases who had a bladder diameter over 15 mm. Our case had a 12 mm bladder in the 14th week of pregnancy, and as previously stated, chorion villus biopsy was reported as Trisomy 18. This is similar with the literature.

If a megacyst is concomitant with an increased nuchal translucency measurement, a 75% chance of chromosomal abnormality has been reported.[8] On the contrary, our case had a nuchal translucency measurement of 1.2 mm.

If a chromosomal abnormality has not been detected in fetuses with bladder measurements between 7-15 mm in early pregnancy weeks, 90% of the cases would carry on to remission. Still, all the cases should be followed up to exclude the presence of an obstructive uropathy. As one would know, bladder is made up of smooth muscle fibers, and its autonomic innervation starts after the 13th week of pregnancy. Earlier than this week, there is prominence of connective tissue and non contractile structures in the bladder. 7-15 mm interval is accepted as a critical diameter for the functional development of fetal bladder. 10% will progress to severe obstructive uropathy. A case reported by Stiller was detected with a 10 mm bladder at the 11th week, which has increased to 30 mm in 13th gestational weeks, and bilateral hydronephrosis has been detected to develop.[9] Again, Zimmer et al.[10] reported another case of 13 mm bladder diameter in the 11th gestational week progressing to 30 mm a week later, progression to hydronephrosis was observed.

There are reports of increased concomittance of obstructive uropathy and renal dysplasia if the bladder diameter is over 15 mm.[11] Such disorders frequently can be corrected in the 2nd trimester and renal damage is possibly prevented. But the success rate and outcome of a decompression procedure attempted in earlier pregnancy weeks could not be completely foreseen yet.

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