Fetal Megacystis and Trisomy 18 Association: Case Report

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Abstract

Objective: We aimed fetal megacystis existence, which is frequently seen together chromosomal anomalies, to discuss a case report.

Case: The pregnancy of a patient who is 31 years old and whose pregnancy is detected fetal megacystis in 14 weeks and detected trisomy 18 by amniocentesis which is made in 16 weeks is terminated in Ağrı Maternity and Children Hospital.

Conclusion: In fetal megacystis, it is detected 25 chromosomal defects of fetuses whose fetal bladder longitudinal diamater between 7-15 mm. From this defects the most frequently seen are Trisomy 13 and likely in our case Trisomy 18. 90% of the cases recover not to develop any sequela or adverse effects in chromosomally normal group. In this case report, we discussed a case which is cooperated with fetal megacystis and Trisomy 18.

Keywords: Fetal megacystis, ultrasonography, trisomy 18.

Fetal megasist ve trizomi 18 birlikteliği: Olgu Sunumu

Amaç: Kromozomal anomaliler ile birlikteliği siklikla izlenen fetal megasist varyeti sunumunu amaçladık.


Anahtar Sözcükler: Fetal megasist, ultrasonografi, trizomi 18.

Introduction

Fetal megasistis which is >7 mm of fetal bladder longitudinal diameter is seen in 1/5000 births. Bladder is an organ in abdomen which is diagnosed simply and fastly with stomach. The routine anomaly screening of pregnancy between 18-20 weeks in 15-20 minutes, the bladder is always seen in all cases. A normal fetus mictures regularly but never definitely empty and always contain somehow residue urine.

Fetal megasistis is two main reason. In the first reason, there must be a problem in the urine output, this case is in the men mainly the
result of the false development of the urethra. The spectrum of the anomalies changes complete urethral atresia to urethral valves that of membrane/prosthetic urethra. Bladder obstruction of women is usually the result of the complex defects of the development of the urogenital system and called 'anomalies of the cloacal bed'. Second group is bladder growth due to nonobstructive factors. These are heterogen group because of the underlying complex pathologies. Among of these neuropathic bladder, obstruction of the small intestines due to muscle degeneration, megacystic micro-colon intestinal hyperperistalsis syndrome and Prune Belly syndrome.

Case

There isn’t an of the patient who first in 14. weeks in 31 years gravida 5, parity 4, live 4. There isn’t an in family anamnesis and laboratory tests were normal. The fetal bladder longitudinal diameter was 19mm and amniotic fluid was normal in the obstetric ultrasonography (Picture 1). NT measurement was 3.2mm. There isn’t another finding in ultrasonography. Because of the detection of the same results of the patient’s ultrasonographic examination which is made in 16 gestational weeks, amniocentesis process is performed. Because of the result of the amniocentesis Trisomy 18, the pregnancy of the patient is terminated by getting permission of the family.

Discussion

In the %25 of the fetuses whose bladder longitudinal diameter is 7-15mm in 10-14 weeks of the pregnancy, it is detected chromosomal defects. Among of these defects the most seen are Trisomy 13 and like our case Trisomy 18. In the chromosomally normal group 90 percent of the cases is improved not to develop any sequela or adverse effect. In the contrary, in the cases of the bladder diameter>15mm the chromosomal anomaly ratio is 10%; but the chromosomally normal cases in this group always together progressive obstructive uropathy. The treatment of the megasistis changes depend of the underlying pathology. If the megasistis develop in the early period of the pregnancy, usually the cause is urethral atresia and these cases are fatal. The being of the obstruction partial or complete effect the treatment. In the detection of the obstruction the best method is measuring of the amniotic fluid level. The obstruction is getting increase the amniotic fluid miktari is getting decrease. In the cases of

![Picture 1. Appearance of the fetus.](image-url)
the oligohydramnios the bladder is emptied by making vesico-amniotic shunt. This perform can be make 3-4 times. It is protected from pulmonary hypoplasia which develops in the cases of the oligohydramnios. In the group of the amniotic fluid level is normal, the treatment of the can be make. Assessing the neuropathic causes of the bladder distension is is harder and the worth of the inuetro treatment of this causes isn’t and it is needed long time randomised studies in this topic.14 Megasistis is in the %75 of the chromosomally abnormal cases and in the %30 of the chromosomally normal cases along with the increased NT. The underlying mechanism of the increasing NT in the fetal megasit is may be thoracic compression.5 In the cases of megasitis which bladder 7-15mm, if the fetal caryotype is normal it is told to parents megasit will improve in the 90% percent of the cases not to cause any adverse effect of kidney development and function. Because of the bladder smooth muscles and autonomic innervation develop after the 13.week; in the pregnancies less than 13 weeks bladder wall is connective tissue no epithelium and contractile element. For this reason, the assesment of this group patients is left to 14. weeks which the bladder completely develop.

**Conclusion**

In fetal megasitis, it is detected 25% chromosomal defects of fetuses whose fetal bladder longitudinal diamater between 7-15mm. From this defects the most frequently seen are Trisomy 13 and likely in our case Trisomy 18. 90% of the cases recover not to develop any sequela or adverse effects in chromosomally normal group. In this case report, we discussed a case which is cooperated with fetal megasitis and Trisomy 18.

**References**