

Fetal Megacystis and Trisomy 18 Association: Case Report

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Abstract

Objective: We aimed fetal megacystis existance, 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 diameter 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 sıklıkla izlenen fetal megasist varlığı sunumunu amaçladık.

Olgu: Ağrı Kadın Doğum ve Çocuk Hastalıkları Hastanesinde otuzbir yaşında ve gebeliğinin 14. haftasında fetal megasist saptanan bir olgunun 16. gestasyonel haftada yapılan amniosentez sonucu Trizomi 18 olarak saptanması üzerine hastanın gebeliği Ağrı Kadın Doğum ve Çocuk Hastalıkları Hastanesinde termine edildi.

Sonuç: Megasist, gebeliğin 10-14.haftasındaki fetal mesane longitudinal çapı 7-15 mm üzerinde olan fetusların %25inde kromozomal defektler saptanmaktadır. Bu defektlerden en sık görülenleri Trisomi 13 ve bizim olgumuzda olduğu gibi trisomi 18'dir. Kromozomal olarak normal olan grupta sonrasında herhangi bir sekel ya da yan etki gelişmeksizin olguların %90'ında spontan düzelme saptanmaktadır. Megasist ve kalın NT nedeniyle amniosentezde uygulanan ve trizomi 18 tanısı konulan olguyu sunmayı 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.¹ 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.²

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 membranous/prosthetic urethra.³ 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 microcolon intestinal hyperperistalsis syndrome and Prune Belly syndrome.⁴

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 anemnesis and laboratory tests were normal.The fetal bladder longitudinal diamater 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, amnio-

centesis 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 diamater 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 diamater>15mm the chromosomal anomaly ratio is 10%; but the chromosomally normal cases in this group always together progressive obstructive uropathy.⁵ 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 miktarı is getting decrease. In the cases of



Picture 1. Appearance of the fetus.

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 harder and the worth of the inutero treatment of this causes isn't and it is needed long time randomised studies in this topic.^{1,6} Megacystis 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 megacystis may be thoracic compression.⁵ In the cases of megacystis which bladder 7-15mm, if the fetal caryotype is normal it is told to parents megacystis 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 megacystis, it is detected 25% chromosomal defects of fetuses whose fetal bladder longitudinal diameter 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 megacystis and Trisomy 18.

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