Fetal Megacystis Associated with Triploidy: A Case Diagnosed at 14th Week of Pregnancy

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Abstract

Background: The aim of this study was to present the case of early fetal megacystis and triploidy. Fetal megacystis caused by triploidy has rarely been described.

Case: The fetal longitudinal bladder diameter was measured 34 mm at 14 th gestational age by ultrasonography. The diagnosis was confirmed by chorionic villus sampling. The result was reported as triploidy 69 XXY. The fetal termination was performed.

Result: The fetal megasistis associated with triploidy is uncommon. Only one case in the literature has reported megacystis with triploidy. We wanted to add a new case to the literature.

Keywords: Fetal megacystis, triploidy, early gestation.

Fetal megasistis ve triploidi: 14. gebelik haftasında saptanan bir olgu

Amaç: Bu çalışmanın amacı erken gebelik haftasında ortaya çıkan fetal megasistis ile birliktelik gösteren triploidi olgusunu sunmaktı. Triploidi kaynaklı fetal megasistis oldukça nadir görülen bir durumdur.

Olgu: Hastanın 14. gebelik haftasında yapılan ultrasonografik incelemesinde longitudinal mesane çapı 34 mm olarak ölçüldü. Tanı koryonik villüs örneklemesi ile doğrulandı. Olguda fetal karyotipleme sonucu 69 XXY (Triploidi) tespit edildi. Terminasyon işlemi uygulandı.

Sonuç: Fetal megasistis olgularında fetal triploidi birlikteliği oldukça nadirdir. Bizim erişebildiğimiz mevcut literatürde fetal megasistis olgularında bir adet triploidi olgusu bildirilmiştir. Biz literatüre yeni bir olgu eklemek istedik.

Anahtar Sözcükler: Fetal megasistis, triploidi, erken gebelik.

Background

Fetal megacystis is defined as expanded bladder higher than normal limits in any gestational period. While it appears as a temporary phenomenon in some cases, it can be an early indicator of lower urinary system obstruction.¹ While megacystis diagnosis is assessed subjectively at second and third trimester, longitudinal bladder diameter about 7 mm and over in first trimester (10th-14th gestational week) is defined as fetal megacystis.² 811 mm bladder diameter is classified as Grade 1 (light), between 12-15 mm is classified as Grade 2 (medium), and over 15 mm is classified as Grade 3 (severe) in first trimester. While many rates were reported in different studies about megacystis prevalence within first trimester, Sebire et al found 15 (1/1633) cases with fetal megacystis in their study in which they examined 24.492 ultrasonography examinations.² In this article, we presented a rare case who had megacystis and triploidy together.

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Case

Our case was found during a routine antenatal observation in an association which is a university hospital and training center. Our case was 25 years old and her husband was 33 years old. Her gravida was 2 and her parity was 0. The patient had D&C history applied due to one year ago. There was no consanguineous marriage between individuals. Gestational sac with 199 mm large in diameter was found in ultrasonographic examination performed on the 6th gestational week of the patient. Nuchal translucency was found as 1.6 mm in the ultrasonographic examination performed on the 11th gestational week. Ultrasonographic diagnoses compatible with gestational week were observed. Fetal megacystis was found in ultrasonographic examination performed on the 14th gestational week. Longitudinal bladder diameter was measured as 34 mm (Figures 1 and 2).

The patient was examined in high risky gestation unit. After taking her informed consent, chorionic villus sampling (CVS) was applied. Afterwards, gestation termination was performed by misoprostol 200 mcg / 6 hours protocol by taking informed consent of the patient (cytotec 200 mcg blister, by vaginal way). The termination process was completed at 36th hour after 6 doses (total dose: 1200 mcg). While the abdomen was observed as distended in the macroscopic examination of fetus, pathological examination was evaluated as urethral atresia. As a result of culture from chorionic villus sampling, karyotyping of fetus was reported as 69 XXY (Triploidy). Maternal and paternal karyotyping was evaluated as normal.

Discussion

Chromosomal defect rate was reported as 21% in the current literature for rarely seen fetal megacystis cases. While trisomy 13 was the most frequent among current chromosomal anomalies, triploidy was observed rarely. Within chromosomal studies performed on fetal megacystis cases in literature, Sebire et al found chromosomal defect only in 3 of 15 cases.² In these cases, Trisomy 13



Figure 1. Fetal megacystis: longitudinal appearance of fetal bladder.



Figure 2. Fetal megacystis: transverse appearance of fetal bladder.

was observed in one case, Trisomy 21 in one case and unbalanced translocation was observed as 14/20. Favre et al reported totally 16 cases within their study and while there was no chromosomal defect after karyotyping in isolated megacystis cases, chromosomal defect was found in four cases within those having accompanying other anomalies. Two of them had trisomy 13, one case had trisomy 21 and one case had trisomy 18.3 Liao et al found chromosomal defect in 30 of 145 cases with fetal megacystis. Trisomy 13 was observed in 17 cases, trisomy 18 in seven cases, trisomy 21 in two cases, trisomy 4 in one case, mosaic trisomy 15 in one case, unbalanced translocation in one case and triploidy was observed only in one case.⁴ While longitudinal bladder diameter was 34 mm in 14th gestational week in our case, chromosomal analysis result was found as triploidy. Its association with fetal triploidy megacystis is very rare.

References

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