

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 14th 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 megacystis 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 megacystis ve triploidi: 14. gebelik haftasında saptanan bir olgu

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

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

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

Anahtar Sözcükler: Fetal megacystis, 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 8-

11 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.

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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