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PRENATAL DIAGNOSIS OF TRISOMI 13: A CASE REPORT

TRIZOMI 13'ÜN PRENATAL TANISI: OLGU SUNUMU

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ÖZET

Bu çalışmada 30 yaşında, iki yıllık infertilite öyküsü olan ve intrauterin inseminasyon sonucu gebe kalan, kromozomal anomaliler açısından prenatal tarama testinin trizomi 21 için yüksek risk gösterdiği ve amniyosentezle elde edilen hücrelerin 15 tanesinin sitogenetik incelemesinde 47,XV, + 13 karyotipi gösterilen bir olgu sunulmuştur. Gebelik 23. haftada misoprostol uygulamasıyla sona erdirilmiştir.

ABSTRACT

We report a 30 years-old primary infertile patient who conceived after intrauterine insemination. The prenatal screening test for chromosomal anomalies showed an increased risk for trisomy 21 at 75 gestational week. Amniotic fluid cytogenetic examination and karyotype of the cells obtained from the umbilical cord confirmed the diagnosis of trisomy 13.*

INTRODUCTION

The incidence of trisomy 13 increases with advanced maternal age. Trisomiy 13 is a clinically significant autosomal trisomy. Its total prevalence is 1,91 per 10.000 births. It results in recognizable patterns of malformations such as ventricular septal defect, urinary tract anomalies, omphalocele, central nervous system abnormalities, cleft palate-lip, microphtalmy and polydactily. Stillbirths and neonatal deaths are common. Stili it is inaccurate to charecterize this disorder as lethal because 38% trisomy 13 children were living at age one year. This decreases to 3-5% by age 5 year. Profound mental retardation is associated with trisomy 13. In vitro fertilization and embryo transfer pregnancies are characterized by an increased-frequency of cytogenetic abnormalities found at prenatal diagnosis. Second trimester maternal serum alfa-fetoprotein, unconjugated estriol, and human chorionic gonadotropine levels are not useful in detecting fetal trisomy 13 and protocols are already existing for Down syndrome or trisomy 18 screening will not detect the majority of cases of this aneuploidy (1).

CASE

A 30-year-old primary infertile patient conceived after intrauterine insemination. She had a history of primary infertility for two years. The prenatal screening test for chromosomal anomalies showed an increased risk for trisomy 21 at the 15th gestational week. The ultrasonografic evaluation of the fetus including the amniotic fluid index and the nuchal translucency (NT) was in normal

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range at same week. Amniotic fluid cytogenetic examination revealed the karyotype 47,XY, + 13 on 15 cells, no mosaicism was seen. Karyotype of the cells obtained by cordocentesis demonstrated same result with the amniotic fluid cytogenetic examination. The pregnancy was terminated by the misoprostol administration at 23rd gestational week. Macroscopic evaluation of the fetus showed bilateral cleft palate-lip (Figüre 1).

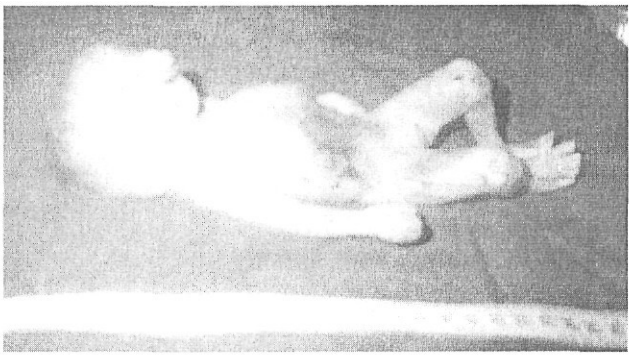

Figure 1. Macroscopic appearance of the fetus showing cleft palate and lip

DISCUSSION

Very few autosomal trisomies survive to birth, the three most common being those for chromosome 13, 18 and 21 giving rise to syndromes Patau, Edwards and Down respectively (2). Trisomy 13 is the third most common trisomy and at 10-14 weeks of gestation the relative proportion of trisomy 21 to trisomy 13 is about eight to one (3). The total prevalence of trisomy 13 is 1,91 per 10.000 births (4).

The birth of an infant with a chromosomal abnormality such as trisomy 18, 13, Wolf-Hirschhorn (4p-) syndrome, Cri-du-chat (5p-) syndrome and the microdeletion syn dromes creates a stressful and devastating experience for families. Many of these disorders have severe consequences encompassing majör malformations and mental retardation (5).

Not only the amniotic fluid cytogenetic examinations and the karyotype of the cells obtained from the umbilical cord but also the rapid florescent in situ hybridization analysis of interphase and high resolution ultrasound examinations are useful to determinate genetic disorders. Feldman et al (6) conclude that the rapid florescent in situ hybridization analysis of interphase, uncultured fetal cells is an accurate and very sensitive method for routine prenatal diagnosis of the most common aneuploidies in high risk pregnancies. İn recent years the measurement of fetal nuchal translucency in the first trimester became a routine screening test for chromosomal abnormalities. Jemmali et al (7) demonstrated that nuchal translucency greater than 2,5 mm between 10 and 14 weeks' gestation is a sonography sign associated with 7,4% of chromosomal anomalies. İn recent years screening of both maternal serum free [3-hCG and pregnancy-associated plasma protein A (PAPP-A) also became popular for the prediction of fetal trisomies in early pregnancy. Spencer et al (8) demonstrated that in trisomy 13 patients at 10-14 weeks of gestation both of this laboratory parameters are decreased and that fetal nuchal translucency is increased. Despite ali these progressions, prenatal diagnosis of trisomy 13 remains possible only by obtaining fetal cells for karyotype analyses. A good screening test with high predictive values is not available at the present time.

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