

# Ultrasound findings in aneuploidy fetusus: Evaluation of 332 cases

## *Anöplidisi olan fetüslerin ultrasonografi bulguları: 332 vakanın değerlendirilmesi*

Ahmet Yalınkaya<sup>1</sup>, Ali İrfan Güzel<sup>1</sup>, Kadir Kangal<sup>1</sup>, Ayşegül Türkyılmaz<sup>2</sup>, Zelar Savaş<sup>1</sup>

<sup>1</sup> Department of Obstetrics and Gynecology, School of Medicine, Dicle University, Diyarbakır, Turkey

<sup>2</sup> Department of Medical Biology and Genetics, School of Medicine, Dicle University, Diyarbakır, Turkey

### Abstract

**Objective:** To evaluate the ultrasound findings found on ultrasound examination among cases that had aneuploidy at amniocentesis.

**Material and Methods:** This prospective study was performed at Dicle University, School of Medicine, Department of Obstetrics and Gynecology. 332 cases applied to our department for prenatal diagnosis and amniocentesis (AC) was performed. Of these cases, twenty were found to have aneuploidy evaluated. The factors recorded were; mean age, gestational weeks, AC indications, ultrasound findings (by Toshiba 140A and GE Voluson 730 Pro 4D ultrasound device) and fetal anomalies.

**Results:** 332 cases have had AC by an experienced specialist, in a two year period. The mean age of the cases was  $32.20 \pm 6.03$  years (22-44), and gestational weeks  $16.45 \pm 1.46$  (13-19). AC indications were; high double and/or triple test with ultrasound findings and abnormal ultrasound findings. In 8 (2.40%) cases there was no reproduction on cell culture. In 14 (4.21%) cases, different types of chromosomal anomalies were detected. In these cases, peripheral blood was taken from the parents and if, at least in one of them this situation was present, this would be accepted as normal. In 20 (6.02%) cases aneuploidy (numerical chromosomal anomalies) were detected and 11 of them (55.00%) were trisomy 21. In all of these aneuploidy cases, different types of ultrasound findings were detected; most of them had multiple ultrasound findings, and some of them had one anomaly. Of all 20 aneuploidy cases; termination of pregnancy was decided in 17 (85%) of them. 3 (15%) of these cases decided to carry on their pregnancy. Of the 3 cases; one baby was delivered spontaneously and live, one had died in utero and labor was induced and the third pregnancy is ongoing.

**Conclusion:** The importance of ultrasound in fetal anomaly screening is incontrovertible and positive ultrasound findings are the most important indications of amniocentesis. For this reason, before amniocentesis, we advise a detailed ultrasound examination by an experienced specialist. (J Turkish-German Gynecol Assoc 2010; 11: 145-8)

**Key words:** Amniocentesis, aneuploidy, ultrasound findings

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### Introduction

Aneuploidies are the most frequent chromosomal anomalies; the incidence was reported as 9 in 1000 live births (1). The most frequent methods used for diagnosing chromosomal abnormalities is karyotyping of fetal cells by invasive procedures such as chorionic villus sampling and amniocentesis (2). Down syndrome, among all aneuploidies, is the most commonly

### Özet

**Amaç:** Amniyosentez ile anöplidisi tanısını almış olan vakaların ultrasonografi bulgularının gözden geçirilmesi.

**Gereç ve Yöntemler:** Bu prospektif çalışma Dicle Üniversitesi Tıp fakültesi Kadın hastalıkları ve Doğum bölümünde gerçekleştirildi. Prenatal tanı ünitemize gelen ve amniyosentez yapılmış 332 vaka ele alındı. Bunlardan 20 vakada anöplidisi saptandı. Yaş, gebelik haftası, amniyosentez endikasyonları, ultrason bulguları (Toshiba 140A and GE Voluson 730 Pro 4D ultrason cihazı ile) ve fetal anomaliler kaydedildi.

**Bulgular:** Bu 332 amniyosentez 2 yıl süresince deneyimli bir hekim tarafından yapıldı. vakaların ortalama yaşı  $32.20 \pm 6.03$  yıl (22-44) ve gebelik haftaları  $16.45 \pm 1.46$  (13-19) idi. Amniyosentez endikasyonları; yüksek ikili ve/veya üçlü test ile pozitif ultrasonografi bulguları idi. Sekiz (%2.4) vakada kültürde hücre üretilmedi. Ondört vakada (%4.2), değişik tip kromozom anomalileri saptandı. Bu vakalarda ebeveynlerden periferik kan kültürü yapıldı ve en az birinde bu varsa bu durum normal olarak değerlendirildi. Yirmi (%6.0) vakada anöplidisi saptandı. Bunların 11 tanesi ise (%55.0) trizomi 21 idi. Tüm bu anöplidisi vakalarında değişik ultrason bulguları tespit edilmişti. bu vakaların 17 tanesi (%85) terminasyonu kabul etti, 3 (%15) tanesi ise gebeliğin devamına karar verdi. Bu gebeliklerin ikisi doğum ile sonlandı, bir bebek yaşıyor diğeri ise kaybedildi.

**Sonuç:** Fetal anomali taramasında ultrasonografinin rolü tartışılmazdır ve pozitif ultrason bulgusu amniyosentezin en önemli endikasyonlarını oluşturur. Bu nedenle amniyosentez öncesi deneyimli bir uzman tarafından detaylı ultrason incelemesini öneriyoruz.

(J Turkish-German Gynecol Assoc 2010; 11: 145-8)

**Anahtar kelimeler:** Amniyosentez, anöplidisi, ultrason bulguları

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encountered chromosomal abnormality in newborns, with an incidence of 1 in 800 live births. Because of decreased mentality and congenital heart disease, Down syndrome, therefore, has a substantial socioeconomic impact (3, 4). With a detailed ultrasound examination by an experienced specialist before invasive prenatal testing, many fetal anomalies may be detected. Therefore, this analysis is called genetic sonography (5).

The aim of this study was to evaluate the findings detected on ultrasound examination for prenatal diagnosis in the cases where amniocentesis was performed.

### Material and Methods

This prospective study was performed on 332 cases who applied to our department for prenatal diagnosis and amniocentesis (AC), the findings of 20 cases having aneuploidy evaluated, from January 2007 to December 2008, at Dicle University, School of Medicine, Department of Obstetrics and Gynecology. This is a reference hospital in the South Eastern region of Turkey. Most of the patients applying to our department have a lower socioeconomic status and the ratio of interrelative marriages is higher than the other regions of Turkey.

The factors recorded were; mean age, gestational weeks, gravidity and parity of the cases. The ultrasound examination and invasive testing were made by a single experienced specialist. The ultrasound findings were recorded on the amniocentesis form.

Indication for amniocentesis, ultrasound findings, fetal anomalies, karyotype results and treatments were also evaluated. The genetic sonogram was made by our Genetic Department after the AC.

### Results

During the study period, 332 cases that had AC for prenatal diagnosis by an experienced specialist were included in the present

study. The mean age of the cases was  $32.20 \pm 6.03$  years (22-44) and gestational weeks  $16.45 \pm 1.46$  (13-19). AC indications were; high double (n=5, 25%) or triple test (n=4, 20%) with ultrasound findings and abnormal ultrasound findings (n=8, 40) (Table 1). In 8 (2.40%) cases, there was no reproduction on cell culture. In 14 (4.21%) cases, different types of chromosomal anomalies were detected. In these cases, peripheral blood was taken from the parents and if, at least in one of them this situation was present, this would be accepted as normal. In 20 (6.02%) cases aneuploidy (numerical chromosomal anomalies) was detected and 11 of them (55.00%) were trisomy 21 (Figure 1).

In all of the aneuploidy cases, different types of ultrasound findings were detected; most of them had multiple ultrasound findings, and some had one anomaly. Of all 20 aneuploidy cases; ,termination of pregnancy were decided in 17 (85%) of them, 3 (15%) of these cases decided to carry on with their pregnancy. Of the 3 cases; one baby was delivered spontaneously and live, one died in utero and labor was induced and one pregnancy is still ongoing (Table 2).

### Discussion

Prenatal diagnosis for fetal aneuploidy began in the 1960's only considering the mother age (5). Later, the American College of Obstetricians and Gynecologists (ACOG) advised prenatal screening for cases 35 years and over (6). With only an advanced maternal age indication only 30% of the Down

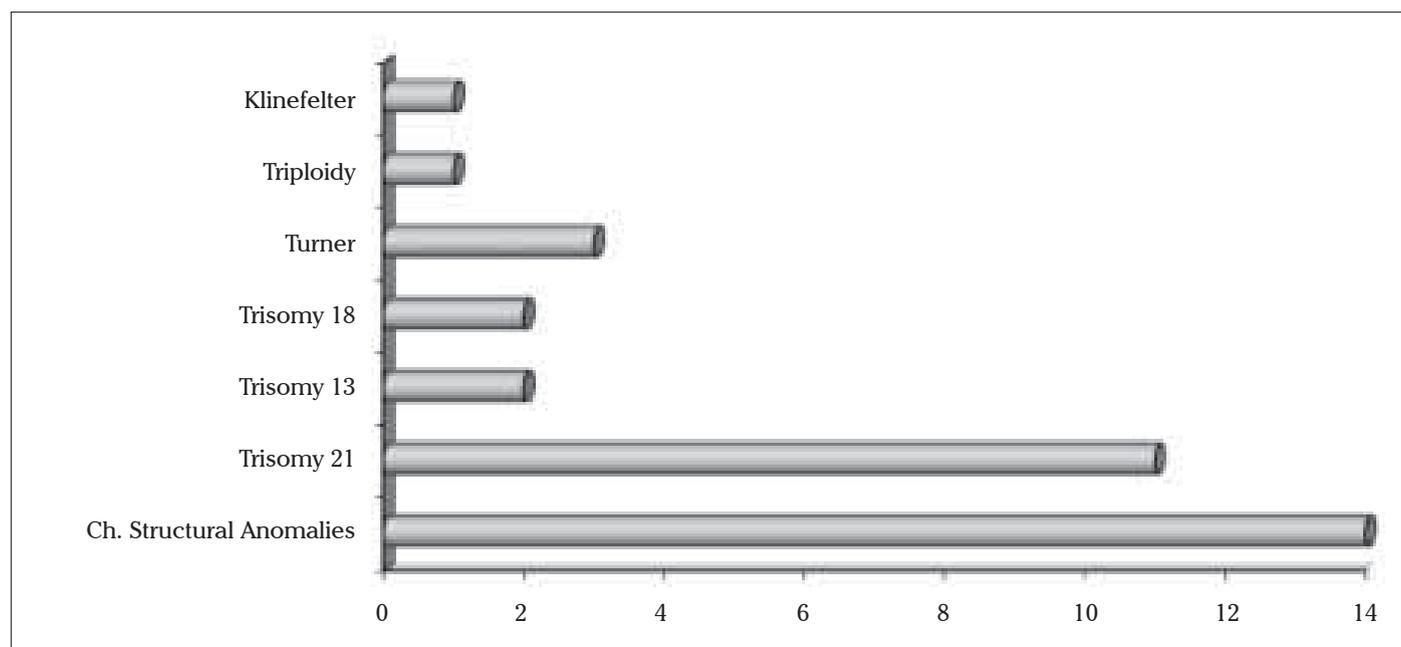
**Table 1. Demographic characteristics and amniocentesis indications of the aneuploidy cases**

Age	Karyotype	G	P	A	Y	GW	Amniocentesis Indication
29	Klinefelter	3	2	0	2	17	Ultrasound findings
28	Triploidy	1	0	0	0	15	Ultrasound findings, high triple test
41	Trisomy 13	14	11	2	10	17	Ultrasound findings, maternal age
34	Trisomy 13	4	2	1	2	17	Ultrasound findings, high double test
37	Trisomy 18	8	7	0	7	16	Ultrasound findings
25	Trisomy 18	3	2	0	1	17	Ultrasound findings
27	Trisomy 21	2	1	0	1	16	Ultrasound findings, high triple test
32	Trisomy 21	2	1	0	1	17	Ultrasound findings, high double test
35	Trisomy 21	2	1	0	1	16	Ultrasound findings, high double test
38	Trisomy 21	4	3	0	3	16	Ultrasound findings, hightriple test
37	Trisomy 21	5	3	1	3	13	Ultrasound findings
35	Trisomy 21	5	3	1	3	17	Ultrasound findings
39	Trisomy 21	4	3	0	3	18	Ultrasound findings, maternal age
31	Trisomy 21	1	0	0	0	17	Ultrasound findings, high double test
32	Trisomy 21	2	1	0	1	17	Ultrasound findings, high double test
44	Trisomy 21	4	2	1	2	17	Ultrasound findings, maternal age
26	Trisomy 21	2	0	1	0	18	Ultrasound findings, high triple test
22	Turner	2	1	0	0	16	Ultrasound findings
25	Turner	1	0	0	0	19	Ultrasound findings
27	Turner	1	0	0	0	13	Ultrasound findings

**Table 2. Ultrasound findings and prognosis of 20 aneuploidy cases**

Karyotype	Ultrasound Findings	Prognosis
Klinefelter	Oligohydramnios, hyperecogenic bowel	Termination
Triploidy	CCA , ventriculomegaly, symmetric IUGR	Termination
Trizomy 13	Echogenic focus in the heart, VSD, hydrocephaly, CCA, talipes, omphalocele, spina bifida, dilated right heart	Termination
Trisomy 13	VSD, pistol shot on hand, left CPC, cryptorchidism	Termination
Trisomy 18	Cystic hygroma, omphalocele	Termination
Trisomy 18	Calvarium ossification defect, hypodensity in all bones, dysplasia of skeleton, achondroplasia	Termination
Trisomy 21	Thick NT, aplasic NB, short extremities	Living
Trisomy 21	Hypoplastic NB, Echogenic focus in the heart	Termination
Trisomy 21	Bilateral CPC, short extremities	Termination
Trisomy 21	Cystic hygroma, hyperecogenic bowel	Termination
Trisomy 21	Anasarca edema, hyperecogenic bowel, cystic hygroma	Termination
Trisomy 21	Cystic hygroma, Anasarca edema, bilateral pyelectasia, aplasic NB	Termination
Trisomy 21	Short extremities, VSD	IUMF
Trisomy 21	Thick NT, aplasic NB, short extremities	Termination
Trisomy 21	Hyperecogenic bowel, Echogenic focus in the heart, Thick NF, hypoplastic NB	Termination
Trisomy 21	Hyperecogenic bowel, left CPC, bilateral pyelectasia, thick NF hypoplastic NB, Face angel : 95°	Termination
Trisomy 21	Hypoplastic NB, Thick NF, Face angel 101°	Pregnancy going on
Turner	Cystic hygroma	Termination
Turner	Cystic hygroma, short extremities	Termination
Turner	Cystic hygroma	Termination

IUGR: Intrauterine growth restriction, CCA: Corpus callosum agenesis, VSD: Ventricular septal defect, NT: nuchal translucency, NB: Nasal bone, IUMF: Intra uterine death of fetus, NF: nuchal fold



**Figure 1. The cases that structural and numerical chromosomal anomalies detected on amniocentesis**

syndromes can be detected (7). Hence, this indication lost its importance.

Nowadays, the main indications are advanced maternal age, biochemical tests and positive ultrasound findings (6). The importance of ultrasound examination also named genetic sonography, has been reported in many studies. The main indication of genetic sonographic examination is the detection of fetal anomalies (8, 9). The risk of fetal aneuploidy increases with the detection of soft markers. The risk increases more with the number of the soft markers (10, 11).

Genetic sonogram has some difficulties such as maternal obesity. Tsai et al. reported in their study that the detection rates of abnormalities on ultrasound screening related inversely with maternal obesity (12)

Nyberg et al. reported that most soft markers in aneuploidy cases were hyperecogenic bowel, ventriculomegaly and cardiovascular system malformations (13). In most of our cases the anomalies were similar to Nyberg's study (Table 2). Of our 20 aneuploidy cases, 7 (14%) were over 35 years age, and advanced maternal age was an amniocentesis indication in only 3 (6%) of them. Chelli et al found that first trimester ultrasound screening may allow early detection of a large number of aneuploidies and fetal malformations (13). In 9 (18%) of the cases, the indication was a high double or triple test. However, in all of the 20 cases, there were ultrasound findings. We consider that this higher ratio of our study is related to higher frequency of the intermarriages and the experience of the specialist who performed the prenatal diagnosis.

In conclusion, nowadays, the importance of ultrasound in fetal anomaly screening is incontrovertible and positive ultrasound findings are the most important indications of amniocentesis. For this reason, before amniocentesis we advise detailed ultrasound examination by an experienced specialist.

#### Conflict of interest

No conflict of interest is declared by authors.

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