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Retrospective analysis of 1429 cases who underwent amniocentesis and cordocentesis

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Abstract

Objective: In our clinic, 1429 patients who underwent amniocentesis and cordocentesis for prenatal diagnosis were evaluated retrospectively.

Methods: The cell culture success, prenatal diagnosis indications, detected chromosomal anomalies and the distribution of chromosomal anomalies according to age were studied in 1429 patients who underwent amniocentesis and cordocentesis between 2008 and 2014 retrospectively. Data were analyzed by SPSS 20.0 software.

Results: Overall culture success rate was 95%. In our study, advanced maternal age (n = 577, 40.4%) and increased risk of maternal serum screening (n=556, 38.9%) were the most common two indications. The most common chromosomal anomaly was trisomy 21 (n=39, 62.9%). There was no difference in the frequency of chromosomal anomalies between women under the age of 35 and women over the age of 35 (p= 1.0).

Conclusion: Chromosomal anomaly rate was found to be 4.3% in our study. In this study, it was shown that maternal advanced age is the most common indication of prenatal diagnosis. However, there was no difference in the frequency of chromosomal anomalies between women under the age of 35 and women over the age of 35.

Keywords: Amniocentesis, cordocentesis, chromosome anomaly.

Özet: Amniyosentez ve kordosentez yapılan 1429 olgunun retrospektif analizi

Amaç: Kliniğimizde prenatal tanı amacıyla amniyosentez ve kordosentez uygulanan 1429 hasta retrospektif olarak değerlendirildi.

Yöntem: 2008-2014 tarihleri arasında amniyosentez ve kordosentez uygulanan 1429 hastada hücre kültür başarısı, prenatal tanı endikasyonları, tespit edilen kromozom anomalileri ve yaşlara göre kromozom anomalilerinin dağılımı retrospektif olarak incelendi. Veriler SPSS 20.0 programı ile değerlendirildi.

Bulgular: Tüm olgularda elde ettiğimiz kültür başarısı %95 idi. Çalışmamızda ileri anne yaşı (n=577, %40.4) ve anne serum taramalarında artmış risk (n=556, %38.9) en sık görülen iki endikasyonu oluşturdu. En sık görülen kromozom anomalisi trizomi 21 (n= 39, %62.9) idi. Herhangi bir endikasyonla prenatal tanı yapılan 35 yaş altı kadınlar ile 35 yaş üstü kadınlar arasında kromozom anomalisi sıklığı açısından bir fark bulunamadı (p=1.0).

Sonuç: Çalışmamızda kromozomal anomali oranı %4.3 olarak bulundu. Bu çalışmada, en sık prenatal tanı endikasyonu izole ileri anne yaşı idi. Herhangi bir endikasyonla prenatal tanı yapılan 35 yaş altı grubu ile 35 yaş üzeri izole ileri anne yaşı grubunda kromozomal anomali sıklığının aynı çıktığı görüldü.

Anahtar sözcükler: Amniyosentez, kordosentez, kromozom anomalisi.

Introduction

The rate of major congenital anomalies is between 2% and 4% in babies born alive. The reason in 10% of these babies with anomaly is chromosomal anomalies.[1] Prenatal diagnosis involves the detection of these diseases, of which some display genetic transmission in the early pregnancy period. One of the aims of prenatal diagnosis is to reveal whether there is any genetic disease

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in the fetus at risk during the period which is appropriate for termination in terms of ethics.[2] Today, despite the efficiency of ultrasonography used for prenatal diagnosis, the need for invasive tests continues. Amniocentesis (AC) and cordocentesis (CC) are the two methods commonly used for prenatal diagnosis today's medicine.

The frequent ultrasonographic evaluations and some biochemical tests recently used increase the need for these two methods. Fetal loss rate after amniocentesis is shown as 1/300 – 1/500 and after cordocentesis as 1.4%.[3,4] In this retrospective study, we aimed to evaluate second trimester amniocentesis and cordocentesis procedures carried out in our university within six years for prenatal diagnosis, and to report their results.

Methods

A total of 1429 pregnant women who referred to our clinic between January 2008 and January 2014 and underwent amniocentesis or cordocentesis due to any indication were included to this study. Patient data were obtained from patient files. All pregnant women and their spouses were informed about the procedure and possible complications, risk evaluation was done and written approval forms were signed by all couples who accepted the procedure. All pregnant women were evaluated in terms of Rh incompatibility before the procedure and were applied anti-D Immunoglobulin required after the procedure. Detailed ultrasonographic evaluation was carried out for all the pregnant women before the procedure. Also, the placental localization and fetus position were evaluated by ultrasonography before the procedure and the most appropriate location was planned for amniocentesis or cordocentesis. Transplacental transition was avoided during amniocentesis. Sterile gauze bandage to be used before the procedure, two 10 ml and one 2.5 ml sterile disposable injectors for amniocentesis, one 2.5 ml sterile disposable injector for cordocentesis and 22 gauge spinal needle for both procedures were prepared on a sterile cover. Amniocentesis was applied between 16 and 20 weeks of gestation and cordocentesis was applied between 20 and 26 weeks of gestation together with ultrasonography in accordance with the known rules. In amniocentesis procedure, 2 ml amniotic fluid aspirated into a separate injector in order to decrease maternal contamination risk. Cordocentesis was carried out from the entrance point of cord to placenta or from free cord depending on the placenta localization by drawing 2 cc fetal blood into the injector already including 0.5 cc heparin. All pregnant women were shown the fetal heart beats after the procedure.

Samples collected for karyotyping analysis were cultured for 3 days for cordocentesis and for 15-20 days for amniocentesis by the methods suitable for the samples, and culture extractions were done. Giemsa banding technique was used. In all cases, 25 metaphase plates were evaluated for structural irregularities and 50 metaphase plates were evaluated for numerical irregularities. Computerized analysis system was used in karyotyping analysis.

Statistics

Data were given as mean and percentage. Significancy of two percentages was evaluated by chi square test. Data was entered to SPSS 20.0 (SPSS Inc., Chicago, IL, USA). $P<0.05$ was considered as significant.

Results

During six years, amniocentesis and cordocentesis were applied to 1429 patients. Cordocentesis was applied to 252 (17.6%) of the patients and amniocentesis was applied to 1177 (82.4%) patients. The ages of our patients ranged between 16 and 51 and mean age was 32.5. Five hundred and seventy-seven (40.3%) patients were 35 years old and above. While mean week of gestation was 17 weeks and 4 days in amniocentesis patients, it was 21 weeks and 6 days in cordocentesis patients. Overall cell culture success was 95 (1358/1429).

The indication groups for prenatal diagnosis were determined as advanced maternal age (35-year-old and above), increased risk at maternal serum screenings (threshold value at double and triple test: 1/270), patho-

Table 1. Prenatal diagnosis indications.

Prenatal diagnosis indication	n	%
Advanced maternal age	577	40.4
Increased risk at maternal serum screenings	556	38.9
Pathological ultrasonography findings	247	17.3
Poor obstetric history	28	1.9
Other	21	1.5
Total	**1429**	**100**

logic ultrasonographic finding, poor obstetric history and others (history of delivering baby with anomaly, IUGR, maternal anxiety, history of delivering baby with chromosomal anomaly, intrauterine transfusion, Rh incompatibility). Our most common indication was advanced maternal age (40.4%) followed by increased risk at maternal serum screenings (38.9%) (**Table 1**).

Chromosomal anomaly was found in 62 (4.3%) patients. The most common chromosomal anomaly was trisomy 21 (n=39, 62.9%) (**Table 2**). Normal variants were found in 46 (3.2%) patients.

According to age groups, chromosomal anomaly was found in 10 (16.1%) patients who were 16- to 25-year-old, in 27 (43.6%) patients who were 26- to 34-year-old, in 15 (24.2%) patients who were 35- to 40-year-old and 10 (16.1%) patients who were 41-year-old and above. In our study, the rates of babies with chromosomal anomaly between women under the age of 35 and women with advanced maternal age over 35 who underwent prenatal diagnosis were found similar (p=1.0) (**Table 3**).

Table 2. Detected chromosomal anomalies.

Karyotype*	AC	CC	Total
47, XY, +21 or 47, XX, +21	36	1	37
47, XY, +18 or 47, XX, +18	4	2	6
47, XY, +13 or 47, XX, +13	2	3	5
47, XY, +mar or 47, XX, +mar	3		3
45,XY,der(13;21)(q10;q10) or 45,XX,der(13;21)(q10;q10)	1	1	2
46, XY, der (15;21)(q10;q10), +21, inv (9)(p11;q12)	1		1
46, XY, der(21;22)(q10;q10),+21		1	1
45, XY, der(13;14)(q10;q10)		1	1
46, XX, del(8)(p12->pter)	1		1
46, XY, t(6;22)(p21.3;q13.3)	1		1
46, XY, t(1,5)	1		1
45, X[75] / 46, XY[25]	1		1
46, XY[60] / 46,XX[40]		1	1
46, XX, inv (12)(p11q14)	1		1
	52	10	62
Normal variants	**AC**	**CC**	**Total**
46, XY, inv (9)(p11;q12) or 46, XX, inv (9)(p11;q12)	5		5
46, XY, 1qh+ or 46, XX,1qh+	10	2	12
46, XY, 16qh+ or 46, XX,16qh+	19		19
46, XY, 16qh+, 1qh+	1		1
46, XX, 14ps (+)	1		1
46, XY, 15ps+ or 46,XX,15ps+	4	1	5
46, XX, 15ps+,16qh+	1		1
46, XY, 21ps+	1		1
46, XX, 22 ps+	1		1
	43	3	46
Total	**95**	**13**	**108**

*Karyotpe and normal variants were determined according to ISCN.[5]

Discussion

Prenatal diagnosis is used frequently in obstetrics. Amniocentesis and cordocentesis are the most common methods used for prenatal diagnosis. In these methods for prenatal diagnosis, culture success rates vary among the laboratories. Saatçi et al. reported the rate of AC as %97 and the rate of CC as 93.6%; Kaplan et al. reported the rate of AC as 98.23%; Cengizoğlu et al. reported the rate of AC as 99%.[6-8] In our study, cell culture success was found as 95.03%. Cordocentesis was applied to 17.6% of our patients. The reasons for high cordocentesis rate are the patients referred to our hospital from nearby cities for prenatal diagnosis and the patients who did not referred to our hospital for pregnancy follow-up at early period.

In most of the AC and CC procedures for prenatal diagnosis, the most common indications were advanced maternal age and the increased risk at maternal serum screenings. The rates for these two indications were reported as 36% and 21%, respectively by Saatçi et al., as 53.4% and 22.4%, respectively by Kaplan et al., and as 18.4% and 69.5%, respectively by Han et al. in a study with a crowded population.[6,7,9] In our study, we found the rate of advanced maternal age as 40.37%, and the rate of increased risk at maternal serum screenings as 38.91%. We interpreted these differences between the rates that some patients underwent both advanced maternal age and maternal serum screenings; however, they were listed among the latter. We planned our study by including patients with high maternal serum screen-

Table 3. The rates and percentages of chromosomal anomalies according to the age.

Age group	Patient number	Number /percentage of chromosomal anomaly
16-25	272	10 / %3.7
26-34	569	27 / %4.8
35-40	372	15 / %4
≥41	216	10 / %4.6
16-34	841	37 / %4.4
≥35	588	25 / %4.3

ing into this group after excluding the patients with advanced maternal age. As in all other studies, abnormal ultrasonographic findings followed these two indications in our study. In AC and CC procedures carried out for prenatal diagnosis, chromosomal anomaly incidences were reported between 1.2% and 10%.[10,11] In our study, chromosomal anomaly incidence was 4.34%. The most common chromosomal anomaly among the babies born alive is trisomy 21. The incidence of trisomy 21 at second trimester is higher than the babies born alive. The reason is that some of these babies may be lost as intrauterine due to various reasons as of second trimester.[12] The rate of trisomy 21 which is the most common chromosomal anomaly among the chromosomal anomalies found by prenatal diagnosis was reported by Zhang et al. as 35.6%, by Han et al. as 36.9%, by Ocak et al. as 60.2%. In our study, this rate was found as 62.90%, which is similar to Ocak et al.[9,12,13] It is known that chromosomal anomalies increase together with age. Sjörgen et al. found the rate of chromosomal anomaly as 2.2% in mothers above 35-year-old, and as 5.3% in mothers above 40-year-old.[14] In a thesis study carried out in Turkey, the risk was found to increase 2.08 times when those between 16- and 34-year-old and those 35-year-old and above were compared in terms of chromosomal anomalies.[15] In our study, chromosomal anomalies were found in 4.3% of the patients who undergone prenatal diagnosis procedure due to the indication of advanced maternal age risk above 35-year old, and in 4.4% of the patients who undergone prenatal diagnosis tests due to other indications under 35-year-old except advanced maternal age.

Conclusion

In our study, prenatal diagnosis tests were referred mostly due to advanced maternal age and increased risk at maternal serum screenings and chromosomal anomaly was detected in 4.3% of the pregnant women. This rate indicates the significance of genetic screening. The most common chromosomal anomaly is trisomy 21. The patients, who underwent prenatal diagnosis procedure due to any indication, were excluded from our study. No significant difference was found in terms of chromosomal anomalies between the patients who undergone the procedure due to advanced maternal age above 35-year-old and the patients who undergone the procedure due to other indications below 35-year-old.

Conflicts of Interest: No conflicts declared.

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