

## *The outcomes of prenatal karyotype analysis in amniocentesis and fetal blood sampling*

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### **Abstract**

**Objectives:** The aim of this study was to evaluate of fetal chromosome analysis in amniocentesis and fetal blood sampling cases.

**Method:** We analysis of 649 cases between January 2007- March 2009 in Dicle University Medical Faculty Department of Genetic. The study was included 552 amniocentesis and 97 fetal blood sampling cases. Lymphocyte culture prepared in duplicate and totally ten slides were prepared for each sample. One of the ten slides was stained with direct Giemsa staining and the others were stained with Giemsa Banding Technique (GTG Banding). A total of 6490 (649x10) slides were evaluated for diagnosis.

**Results:** A total of 649 samples were analyzed for chromosome aberrations and 267 (41%) cases had 46 XX, 257 (40 %) cases had 46, XY normal karyotype. Of the cases, 111 (17%) were detected to have abnormality. Due to cells culture failure, the outcomes of 14 (2%) cases were not obtained (10 amniocentesis and 4 fetal blood sampling). The indications of karyotype analysis were 36% with higher triple test risk, 28% with pathologic ultrasound findings, 15% with higher double test risk, 13% with advanced maternal age, 4% with familial diseases history, 2.5% with parental anxiety and 1.5% with bad obstetric anamnesis, respectively. We have no false positive and false negative results in our study.

**Conclusions:** In our study, chromosome aberrations rate (111 (17%)) was found higher than literature, and the most indication was found higher triple tests. We suggested that karyotype analysis should be considered in all high risk patients.

**Keywords:** Amniocentesis, Fetal Blood Sampling, Chromosome Aberrations.

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## **Introduction**

Prenatal diagnosis is one of the important subjects in obstetrics. Anomaly screening, triple screen and genetic amniocentesis are intensively used for this purpose (1,2). Genetic amniocentesis and cordocentesis remain the most common invasive diagnostic procedures for the detection of chromosomal aberrations in fetuses. The means to diagnose the fetal karyotype has provided medical cytogenetics with one of its major areas of application(3).

The frequency of inherited malformations as well as genetic disorders in newborns account for around 3-5%. These frequency is much higher in early stages of pregnancy, because serious malformations and genetic disorders usually lead to spontaneous abortion. Prenatal diagnosis allowed identification of malformations and/or some genetic syndromes in fetuses during the first trimester of pregnancy. A variety of approaches for genetic prenatal analyses are now available, including preimplantation diagnosis, chorion villi sampling, amniocentesis, fetal blood sampling as well as promising experimental procedures (e.g. fetal cell and DNA isolation from maternal blood) (4).

In this study, we investigated fetal chromosome constitution of 649 cases between January 2007-March 2009 in Dicle University Medical Faculty Department of Genetic.

## **Materials and Methods**

The samples were collected from patients who were referred to the Genetic Diagnostic Laboratory of the Department of Medical Biology, Medical Faculty, Dicle University, for prenatal diagnosis during January 2007 to March 2009. A total of 649 samples (552 amniocentesis and 97 fetal blood sampling) were included in this study.

Two Lymphocyte Cultures were prepared for each cases. Karyotype analyses were carried out totally on ten slides for each specimen. One of the ten slides was stained directly with Giemsa staining method and others with Giemsa Banding Technic ( GTG Banding). A total of 6490 (649x10) slides were evaluated for diagnosis. In case of necessity, Molecular cytogenetic technics were applied.

## **Results**

In this study, we evaluated a total of 649 samples which were obtained from pregnant woman during January 2007 to March 2009. A total of 649 samples were analyzed for cytogenetically and 267 (41%) cases had 46 XX, 257 (40 %) cases had 46, XY normal karyotype. Of the cases, 111 (17%) were detected to have abnormality. Due to cells culture failure, the outcomes of 14 (2%) cases were not obtained (10 amniocentesis and 4 fetal blood sampling). The indications of karyotype analysis were 36% with higher triple test risk, 28% with pathologic ultrasound findings, 15% with higher double test risk, 13% with advanced maternal age, 4% with familial diseases history, 2.5% with parental anxiety and 1.5% with bad obstetric anamnesis, respectively. We have no false positive and false negative results in our study.

## Discussion

The practice of Prenatal diagnosis in chromosomal abnormalities has been used for nearly half century. When Steele and Breg showed that the chromosome constitution of a fetus could be determined by analysis of cultured cells from the amniotic fluid in 1966, Prenatal diagnosis had its beginning. Karyotype analysis has been the standart method for prenatal cytogenetic diagnosis since the 1970s (4). Nowadays, a number of different noninvasive tests and genetic prenatal analyses have been developed (1,2,3,4,5).

In Tseng et al. studied a total of 7,028 amniocenteses were performed and analyzed for chromosome aberrations. Among these the most indication was advanced maternal age The highest detection rate of chromosome aberrations was in cases undergoing amniocentesis for abnormal ultrasound findings (8.86%), followed by other reasons (2.74%), abnormal maternal serum screening results (2.60%), and advanced maternal age (2.31%). Chromosome aberrations were detected in 207 cases (2.90%)(3).

In Miyake et al. studied a total of 1063 genetic amniocentesis samples and chromosome aberrations were detected in 35 cases (3.3%)(6).

In Chaabouni et al. studied that 3110 fetal karyotypes carried out in a Tunisian population, by cultured amniocytes analysis. Abnormal karyotypes rate was 4.18%. Common amniocentesis indication was maternal age (7).

In Zhang et al. studied that 2782 cases of high-risk pregnant woman. Chromosomal abnormalities were observed in a 3.99% (111/2782). Abnormal chromosome carriers had the highest percentage of abnormal chromosomes (67.86%) when compared with chromosomal abnormalities in patients with ultra-sonographic "soft markers" (11.81%), advanced maternal age (4.51%)(2).

In our study, chromosome aberrations rate (111 (17%) was found higher than literature, and the most indication was found higher triple tests. We suggested that karyotype analysis should be considered in all high risk patients.

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