Short Communication

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Prenatal Cytogenetic Diagnosis of 1,400 Consecutive Amniocenteses

T.	Thomas	Mathews
D.	Dipesh	Navsaria
R.S.	Ram S.	Verma

Division of Genetics, The Long Island College Hospital, SUNY Health Science Center, Brooklyn, N.Y., USA

Key Words

Amniocentesis
Chromosome
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Abstract

We report on the incidence of chromosomal abnormalities in 1,400 amniocenteses. **Thirty-one** cases (2.21 %) were found to have various types of chromosomal abnormality. The majority of abnormal cases (54.84%) were trisomies. All individuals with trisomic fetuses chose to terminate the pregnancy with the exception of one who gave birth to a Down's syndrome child. However, fetuses with apparently balanced translocations were phenotypically normal at birth.

Dr. Ram S. Verma, Division of Genetics, Long Island College Hospital, Atlantic Avenue at Hicks St., Brooklyn, NY 11201 (USA)

Introduction

A quarter of a century ago, Steel and Breg [1] succeeded in culturing amniotic cells, and obtained the first fetal karyotype. The last two decades have witnessed exponential growth in amniocentesis procedures for ruling out apparent chromosomal abnormalities before birth. The recent advances in the safety and accuracy of the procedure have resulted in a universal acceptance for its routine use in modern medicine [2-5]. We present the incidence of chromosomal abnormalities in 1,400 am-niotic-fluid samples referred to our laboratory.

Materials and Methods

The present study comprises cytogenic findings in 1,400 amniocentesis cases. 10-20 ml of amniotic fluid were collected into 2 sterile plastic centrifuge tubes at a gestational age of 14-17 weeks. The amniocytes were cultured using the in situ technique described earlier [6].

Chromosomes were prepared in the usual manner [7, 8]. The routine diagnosis was performed using the GTG-banding technique.

If the case was found to be cytogenetically abnormal, various selective staining techniques were utilized as appropriate [9]. A minimum of 20 metaphases were examined from each patient. In the case of mosaicism, 50-100 metaphase spreads were analyzed. At least 2 metaphases were karyotyped from each case.

Results and Discussion

Of the 1,400 amniotic fluids referred, approximately 0.5% failed to grow and required retapping. Neither maternal cell contamination nor cytogenetic error was encountered. There was only 1

case with pseudomosaicism, which displayed a normal cell line and another line with a Robertsonian translocation involving chromosomes 15 and 21: the child was cytogenetically normal at birth. Thirty-one cases (2.21 %) were found to have various types of chromosomal abnormalities. The majority of chromosomal abnormalities were autosomal trisomies (54.84%). The incidence of trisomy 21 (38.71 %) was by far the highest. The number of trisomies for chromo-

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somes 13, 18, 21 and 22 were 1, 2, 12 and 2, respectively. Sex chromosome abnormalities were found in 3 cases, while 11 cases were found to have structural aberrations (table 1). To our knowledge, all individuals with trisomic fetuses chose to terminate the pregnancy with the exception of one who carried a Down's syndrome baby to term. However, fetuses with apparently balanced translocations were phenotypically normal at birth. The incidence of chromosomal abnormalities in the general population is estimated to be 0.5 % of live births [10, 11], but within our high-risk population was 2.21 %, 4.44-fold higher than the general population. Obstetricians, epidemiologists and clinical geneticists will find this information useful.

Table 1.

1,400 cases

Prenatal detection of chromosomal abnormalities in

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