SHORT COMMUNICATION

Prenatal cytogenetic analysis in the Presov region (Slovakia) in 1999–2004

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Abstract

Prenatal genetic diagnostics is a part of prenatal care. Prenatal karyotyping is used to identify major genetic and congenital abnormalities in a developing fetus. In the Presov region (Slovakia) in 1999–2004 370 amniotic fluid samples were analysed by G-banding. Abnormal karyotypes were detected in 3.8 % of samples. A karyotype using classical banding methods is the only fully informative method able to detect all chromosomal abnormalities. Identification of fetal abnormal chromosomes in high risk pregnancies allows proper pediatric and obstetric managment of the cases as well as genetic counselling *(Tab. 1, Fig. 2, Ref. 5)*.

Key words: prenatal cytogenetic diagnosis, chromosome aberrations, amniocentesis, karyotyping.

Prenatal diagnosis (PD) is a relatively new branch of medical genetics enjoining presently a rapid practical and scientific progress. It is now routinely offered as a part of prenatal care. Prenatal testing is used to identify major genetic and congenital abnormalities in a developing fetus. Chromosomal abnormalities are responsible for a considerable number of birth defects, and more than 50 % of spontaneous abortions. Chromosomal abnormalities occure in about 0.6 % of all live birth (Sršeň and Sršňová, 2000). All can be diagnosed prenatally, but the risks of invansive prenatal tests may outweigh the benefits.

The aim of the study

The aim of the study was to estimate the incidence of chromosomal abnormalities detected in amniotic fluid samples over the period 1999–2004 in the Presov region. Conventional chromosomal analysis on amniotic fluid samples was performed at the Department of clinical genetic in the Presov region (Slovakia).

Patients and methods

Prenatal diagnosis for detection of chromosomal abnormalities was carried out in risk women for following indications: maternal age 35 years and more, previous child with a chromosomal anomaly, parent with a constitutional chromosomal abnormality and abnormal findings at the ultrasound examinations. Usually either 15–20 ml of amniotic fluid was obtained from each patient depending on the gestation age at the time of sampling. Standard culture and harvest methods, described by Rooney and Czepulkowski (1992) were used to obtain chromosome preparations. The identification and classification of the chromosomes were made based on recommendations of the International System for Cytogenetics Nomenclature (ISCN, 1995).

Results

Over the period of years 1999–2004 prenatal diagnosis was performed in 370 high risk pregnancies presenting at the Department of clinical genetics in the Hospital in Presov.

Chromosomal analysis was successful in 352 instances and uninformative in 18 cases. Culture success varied from 80 % to 100 %. The overall success rate for obtaining cytogenetic diagnosis was 91.7 %. In 18 amniotic fluid samples (4.9 %) no diagnosis could be obtained, mainly due to absence of cell growth in late gestation samples or because of blood contamination. 370 amniotic fluid samples were analysed by G-banding. Normal karyotypes were found in 356 samples (96.2 %). Abnormal karyo-

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Tab. 1. Cytogenetic examination of amniotic fluid in the Presov region.

Year	The number of prenatal cg examinations	Pathological findings		Abnormal karyotype	Cultivate success %
		n	%		
1999	14	-	-	-	100
2000	48	1	2.1	46,XX/46,XX,del (21) (q21-qter)	91.7
2001	70	1	1.4	47,XX,+18	80
2002	62	1	1.6	46,XY(4p-)	90.3
2003				46,XX/46,XX,t(7,20) (q23,p13) 46,XY,inv(9) (p12,q12)	
	94	5	5.3	46,XY,inv(9) (p12,q12)pat 46,XX,22p+ 47,XY,+mar	89.4
2004				46,XX,inv(9) (p12,q12) 46,XY/46,XY,t(5,6) (q25,q21)	
	82	6	7.3	47,XY,+mar 46,XX,inv(9) (p12,q12) 46,XX,inv(9) (p11,q12) 46,XY,inv(9) (p12,q12)	98.8

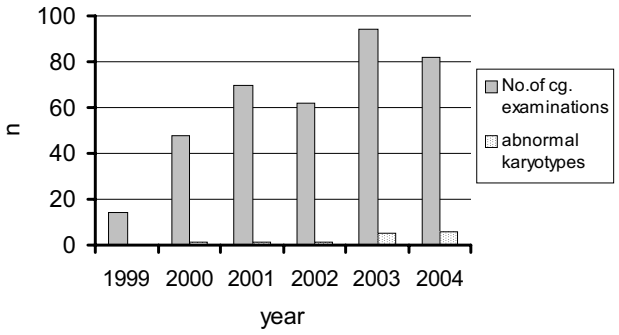

Fig. 1. Cytogenetic analysis of amniotic fluid in the Presov region (1999–2004).

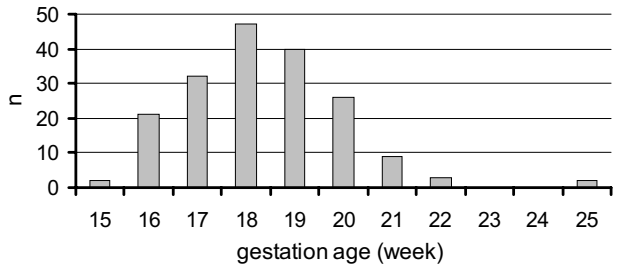

Fig. 2. Distribution of gestation age at the time of amniocenthesis in the Presov region (1999–2004).

type were seen in 14 samples (3.8 %). Generalized mosaicism was found in 3 samples (0.8 %). Pseudomosaicism was detected in 2 (0.5 %) cases. Numerical abnormalities was detected in 3 cases (0.8 %), structurally abnormal karyotypes was detected in 11 cases (3 %). The most common detected chromosomal abnormality was pericentric inversion of chromosome 9 detected in 6 cases (1.6 %). Mosaic karyotype was detected in 3 cases (0.8 %) (Tab. 1). The proportion of abnormal karyotypes that were diagnosed prenatally increased from 2.1 % in 2000 to 7.3 % in 2004 (Fig. 1). Comparing the first years to the last two years the rate of abnormal cytogenetic results increased significantly. The gestational age at the time of amniocentesis was most commonly 18 week. Distribution of gestation age at the time of amniocentesis in the Presov region over a period 1999–2004 is showed in Figure 2.

The present results indicate that amniocentesis is a technically easy and provides results are very acceptable. Cytogenetic diagnostics is the most serious on the time from the all assortment of prenatal examinations. The results of cytogenetic examinations after date 1999–2004 suggest the increasing trend in the proportion of cases that were diagnosed before birth.

Discussion

Prenatal chromosome diagnosis has been a rapidly changing field over the past years for both sampling methodologies and molecular techniques to complet chromosome analysis. Since introduction of cytogenetic analysis in the amniotic fluid cells this method has been considered the standard test for prenatal diagnosis.

Chromosomal aneuploidies especially trisomies 13, 18, 21, monosomy X and 47,XXY account for up to 95 % of live born cytogenetic abnormalities (Kucheria et al, 2002). New methods in prenatal diagnostics allow to demonstrate certain numeric chromosomal aneuploidies in amniotic cells within 24 h in contrast to conventional methods which take 1–3 weeks. Rapid molecular biological methods for prenatal diagnosis of the most common aneuploidies, collectively known as rapid aneuploidy testing are not use as stand-alone test, but some of them are routinely applied as a preliminary test that shorterns the waiting time for classic cytogenetic karyotyping (Dudarewitz et al, 2005).

The present results indicate that amniocentesis is a technically easy and provides results that are very acceptable. Findings from our study and the available evidence in the literature suggest that

prenatal diagnosis might have improved. A karyotype using classical banding methods should be performed whatever the indication of prenatal study is. It is the only fully informative method able to detect all chromosomal abnormalities. The conventional cytogenetic analysis serve as a gold standard at present.

Perspectives of prenatal diagnostics of genetic condition cases with development of alternatives in this department will improve. A big promise to the future is follow-up development of diagnostic by analysis DNA. New technical platforms have enhaced the spectrum of disorder that can be diagnosed prenatally. Prenatal cytogenetic analysis is a standard method for prenatal cytogenetic evaluation. Identification of fetal abnormal chromosomes in high risk pregnancies allows proper pediatric and obstetric management of the cases as well as genetic counselling. Prenatal programme demands connection and cooperation among clinical positions and cytogenetic laboratory.

Progress in clinical managment of genetically determined pathological conditions resulted in a substantial increase in the prenatal diagnosis and a considerable reduction in perinatal mortality of infants.

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