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Genetic Amniocentesis for Prenatal Diagnosis at Bhumibol Adulyadej Hospital: four years experience

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ABSTRACT

Objective To evaluate results of amniocentesis for prenatal diagnosis of chromosome abnormality at Bhumibol Adulyadej Hospital

Design Retrospective descriptive study.

Setting Perinatal unit, Department of Obstetrics and Gynaecology, Bhumibol Adulyadej Hospital.

Subjects Nine hundred twenty patients who had amniocentesis at perinatal unit, Department of Obstetrics and Gynaecology, Bhumibol Adulyadej Hospital, from January 1, 1996 to December 31, 1999.

Results During a four years period nine hundred twenty women underwent genetic amniocentesis for chromosome diagnosis. The most common indication was elderly gravidarum. Other indications were, family chromosome disorder, previous Down syndrome, maternal anxiety and malformed fetus. We found fifteen chromosome abnormalities. One case was contaminated and two cases had tissue culture failure. Most of the amniotic fluid were clear, only seven cases had brownish colour, thirteen cases had blood - stained and six cases had meconium-stained. There were two fetal loss in our study.

Conclusion Genetic amniocentesis were highly successful performed with low complications. Among 920 cases, 15 chromosome abnormalities were detected with this procedure. It is a safe method for prenatal chromosome diagnosis by well-trained operators.

Key words: prenatal diagnosis, amniocentesis, chromosome aberrations

Following the successful culturing of fetal cells from amniotic fluid by Steele and Berg[1] in 1966, mid-second-trimester amniocentesis was established as the safe and accurate standard technique for prenatal diagnosis.[2-4] The procedure is performed on an outpatient basis and is nearly painless. In Thailand, those who are at risk for having a chromosome abnormality fetus will be counselled for mid-second-trimester amniocentesis but not all of them have been done due to inadeguate genetic laboratory and inexperience of operators. In Bhumibol Adulyadej Hospital, we have been performing amniocentesis since 1988. Our cases were increased every year up until 1998. After we developed chorionic villus sampling technique, the number of amniocentesis was slightly decreased. This report analyzed the indications for amniocentesis, the gestational ages, maternal ages and the results of karyotyping.

Materials and Methods

A total of 920 cases of genetic amniocentesis were performed from January 1, 1996 to December 31, 1999. The most common indication for amniocentesis was advanced maternal age. After process of counselling and signing consent form, ultrasound examination was performed with curvilinear probes (Acuson 128xp). Identification of the dividing membrane and placental localization were accomplished. The patient's abdomen was prepped with povidone-iodine and draped. The transducer was covered by a sterile glove and introduced into the field. The transducer was placed to indentify a pocket of amniotic fluid on either side suitable for amniocentesis. A 21-gauge spinal needle was introduced under ultrasound guidance into the amniotic sac. An assistant aspirated amniotic fluid without altering the position of the transducer. The initial 1-2 mL of amniotic fluid was discarded and 20 mL of amniotic fluid was aspirated and sent to laboratory for tissue culture with separated syringe. After the procedure, ultrasound examination was repeated to confirm fetal heart motion.

Results

All of 920 women were performed amniocentesis at our hospital during the study period, 3 cases needed to repeat amniocentesis due to 2 culture failure and 1 bacterial contamination. Most common indication was elderly gravida as shown in Table 1. Ninety-one percent of amniocentesis were done during 16-18 weeks (Table 2). The distribution of the women ages shown in Table 3, 848 (92.17%) were more than 35 year of age. The most common age group were 35-39 years. The youngest age was 20 and the oldest age was 44 as shown in Table 3. Cytogenetic results of chromosomal abnormality were shown in Table 4. The success rate in performing amniocentesis with first puncture was 96.52%. 97.17% of the fluid was clear, 0.76% was brownish colour, 0.65% was meconium-stained and 1.42% had blood-stained. All cases with chromosome abnormalities were terminated except 47 XXY and 46 XX+16P^{+}. There were two fetal loss in our study.

Discussion

The major cytogenetic indications for amniocentesis were advanced maternal age, chromosome abnormality in previous child and family chromosome disorder which contributed about 98.58% of all cases. The minimum maternal age was 20 years and the maximum maternal age was 44 years. The most common maternal age range were 35-39 years. The most common gestational age at amniocentesis was 17 weeks. While we scheduled to perform the procedure between 16-18 weeks. Almost all of the late cases had late booking for antenatal care.

Most of the amniotic fluid was successfully obtained at first attempt (96.52%) and was clear (97.17%). Three cases required second amniocentesis because of two cases had culture failure and one case had contamination. Overall fail cultured rate was 0.32% which was less than other studies (1.74%)[5] Nearly all amniocentesis were performed by well-trained operators. Most of 2 puncture cases were performed by general obstetricians who were not staffs in our perinatal unit. It has been concluded by working group all over the world that genetic amniocentesis is a safe, accurate, reliable with minimal risk if it is performed by an expertise operators.[6] Gerbie an Elias[7] recommended that amniocentesis should be performed by obstetricians and gynecologists 1) who have experience in this procedure, 2) with the availability of high quality ultrasonography and, 3) with access to a laboratory with experience in culturing and analyzing amniotic fluid cells. Finegan and associates found that second trimester amniocentesis does not appear to compromise intelligence, academic achievement, usual-motor-perceptual abilities, fine motor co-ordination, speech articulation, behavior, social competence, growth or health in children at seven years of age[8] whose mother had done this procedure. Complications related to prenatal diagnosis utilizing amniocentesis are intrauterine fetal death,[9] leakage of amniotic fluid[10] direct fetal injury[11] and most commonly miscarriages.[12-14] Fetal loss rates for amniocentesis have ranged from a low of 1.0%[15] to 1.3%.[16] Our study had only 2 fetal loss (0.21%) which was fewer

than previous studies. We found fifteen cases of chromosome abnormality, most of them were from advanced maternal age (93.3%) and the most common cases of chromosome abnormality were trisomy 18 and trisomy 21. Ferguson-Smith and Yates [17] reported all chromosome aberrations in maternal age 35 years and over was about 2.26% which is larger than our study (1.16%).

In conclusion, amniocentesis performed around 16-18 weeks is a safe method for prenatal diagnosis. The risk for pregnancy loss and number of multiple puncture is low in our experience. The diagnosis guides us to the proper management and prevents birth with serious chromosome disorders. For safety reasons, we suggest that amniocentesis should be performed at centers where there are experience operators.

Table 1. Indications for amniocentesis

Indications	No	%
Elderly gravidarum	848	92.17
Family chromosome disorder	30	3.26
Previous Down syndrom	29	3.15
Maternal anxiety	9	0.98
Malformed fetus	4	0.44
		100%

Table 2. Gestationals al ages of the subjects

Gestational ages (weeks)	No	%
16	159	17.28
17	400	43.48
18	279	30.33
19	44	4.78
20	16	1.74
21	22	2.39
		100%

Table 3. Ages and numbers of subject

Ages (years)	No (1996)	No (1997)	No (1998)	No (1999)	Total
20-24	1	1	1	0	3
25-29	3	4	4	3	14
30-34	15	15	8	17	55
35-39	181	215	133	181	710
40-44	38	43	27	30	138
	238	278	173	231	920

Table 4. Abnormal karyotypings ages & indications

No	Types of Abnormality	Maternal ages (years)	Indications
1	47XX+21	40	Elderly
2	46XY, inv(9)(p13;q12)	40	Elderly
3	47XXY	40	Elderly
4	46XX+16P+	40	Elderly
5	47XX+18	39	Elderly
6	46XY,inv(7) (p11;q12)	38	Elderly
7	47XY+18	38	Elderly
8	47XX+18	38	Elderly
9	47XX+21	38	Elderly
10	47XY+13	37	Elderly
11	47XY+21	37	Elderly
12	47XY+18	35	Elderly
13	47XX+18	35	Elderly
14	47XX+21	35	Elderly
15	47XY+18	29	Malformation

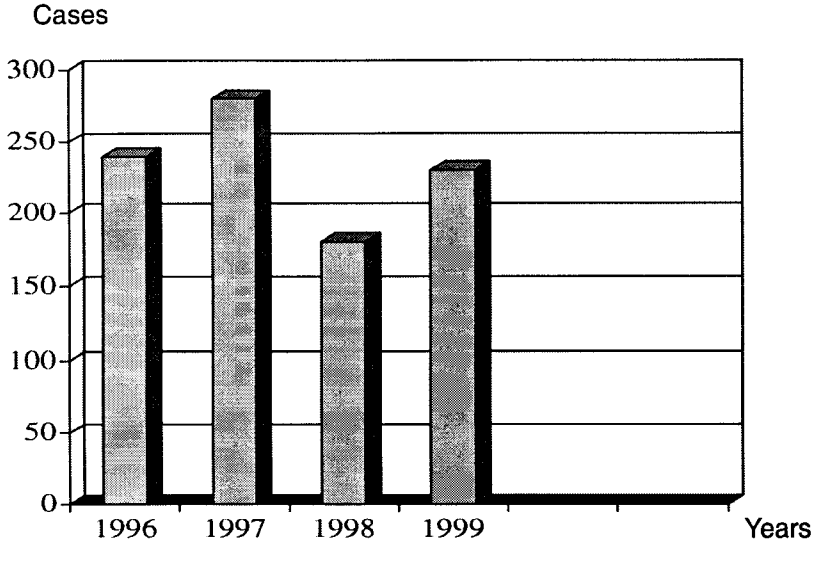

Fig. 1. Total numbers of amniocentesis by years.

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