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Ultrasonographic Fetal Ear Measurement In predicting Fetal Chromosome abnormality

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ABSTRACTS

Objective To study the accuracy of fetal ear measurement in second trimester by ultrasound to predict fetal chromosome abnormalities

Methods Fetal ear lengths were obtained ultrasonographically in 418 singleton pregnancies between 16-19 weeks' gestation in Bhumibol Adulyadej Hospital from January 1, 1997, through September 30, 1999.

Results Linear relationships were found between ear length and gestational age. All abnormal chromosome fetuses had smaller ear length than mean ear length of normal chromosome fetuses between 16-19 weeks.

Conclusions Fetal ear length may be useful in identifying aneuploid fetuses sonographically during the second trimester.

Key words: fetal ear, chromosome abnormality

Recent advances in ultrasound technology have improved the detection of congenital malformations in utero. New approaches for antenatal prediction of chromosomal abnormalities have been suggested.[1-4] It is know that 80% of infants with trisomy 21 are born to mothers under 35 years of age.[5] The addition of maternal triple screening test (alpha-fetoprotein, unconjugated estriol, human chorionic gonadotropin) identifies another 60% of trisomy 21 in mothers under 35 years of age.[6-7] Various sonographic measurements and finding have been proposed to detect these fetuses, including shortened femur length, shortened humerus length, mild renal pyelectasis, hypoechoic bowel and hypoplasia of the middle phalanx of the fifth digit, duodenal atresia, congenital heart disease.[8-15]

Unfortunately each of these methods have deficiencies and most of the abnormal ultrasound findings are inconsistent in an abnormal chromosome fetuses. Cytologic evaluation by amniocentesis, chorionic villus sampling, or fetal blood sampling are still the definitive method for antenatal identification of abnormal chromosome fetuses.

The fetal ear lengths have been reported to be diminished in trisomy 21 fetuses and their potential role in screen for fetal aneuploidy has been suggested.[16,17] However, all the reported studies are based on a small number of cases. We therefore designed an observational study to investigate if the fetal ear length of trisomy 21 is unique and if so, to examine its potential as a prenatal diagnostic test.

Materials and Methods

We studied 418 women with singleton pregnancies undergoing ultrasound examinations for genetic chorionic villus sampling at 10-12 weeks and amniocentesis at 16-19 weeks' gestation between January 1, 1997, through September 30, 1999.

Before amniocentesis or after chorionic villus sampling, each fetus was evaluated sonographically, including standard biometry and a structural fetal survey. Fetal ear measurements were determined by considering the longest longitudinal ear length three times and the average ear length were analyzed. All ultrasound examinations were performed using an Acuson 128XP-10 (Moutain View CA) with a variable-focus 3.5 or 5.0 MHz transducer. Appropriate ear length determinations were obtained by locating the cervical spine longitudinally, and then sliding the transducer tangentially to the temporal bones on either sides of the skull to include the ears in coronal sections.

Statistical analysis were performed by mean and standard deviation.

Table 1. Indications for chromosome studies

Indications	Amnio	CVS	Amnio+CVS	Total
Elderly gravidarum	322	82	2	406
Elevated triple marker	1	-	-	1
Over drugs used during pregnancy	1	-	-	1
Consanguinity	1	-	-	1
Down's syndrome In families	3	-	-	3
Anxiety	5	1	-	6
				418

Table 2. Aged - ratio in chromosome abnormality

Age (years)	No.	Ratio
35	1	1:107
37	1	1:74
38	1	1:57
40	1	1:22

Table 3. Maternal age

Age(years)	Amnio	CVS	Amnio+CVS	Total
20-24	1	-	-	1
25-29	3	-	-	3
30-34	29	4	1	34
35-39	258	73	-	331
40-44	40	6	1	47
>45	2	-	-	2
	333	83	2	418

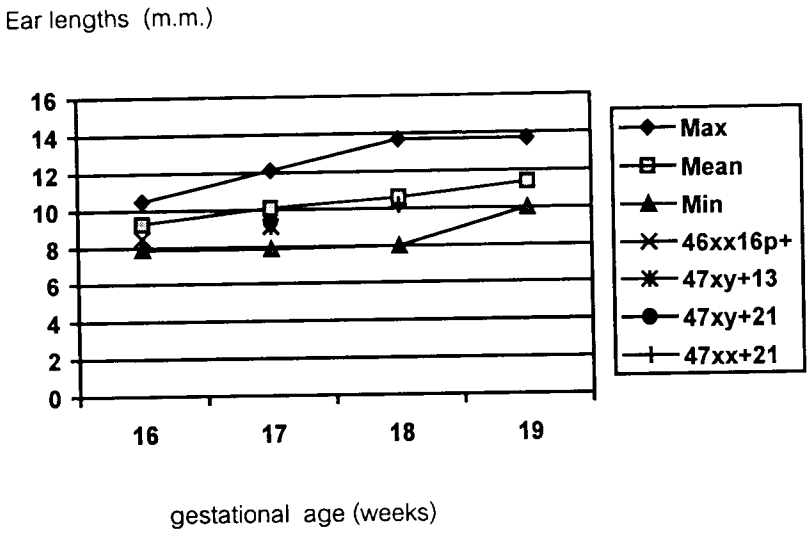

Fig. 1. Demonstration mean ear lengths, Minimum and Maximum ear lengths in normal fetuses, and aneuploid fetal ear lengths.

Results

All of 418 women were included in our study. Most common indication was elderly gravida as shown in Table 1. Aged-ratio in chromosome abnormality was shown in Table 2. Maternal age 35 years old has chromosome abnormality about 1:107 and 40 years old has about 1:22. The distribution of the women ages shown in Table 3,380 (90.9%) were more than 35 years of age. The youngest age was 20 and the oldest age was 46 as shown in Table 3. Mean ear lengths, minimum and maximun ear lengths in normal fetuses, and aneuploid fetal ear lengths was shown in Figure 1.

Discussion

Antenatal karyotyping of fetuses through invasive sampling techniques, e.g. genetic amniocentesis and chorionic villus sampling is not recommended on routine basis due to well-known fetal risks and high costs. Advanced maternal age is not sufficient to define the population at risk as more than 80% of the mothers of Down fetuses are less than 35 years of age.[5] Triple screening test (maternal serum alphafetoprotein, human chorionic gonadotropins and unconjugated estriol) achieved a detection rate of 60% and a false positive rate of 5% in women under 35 years of age.[6-7]

Recently, attention has turned toward finding a fetal marker detectable by ultrasound that would provide a noninvasive, antenatal diagnostic method for aneuploidy.

Abnormally short ears have long been noted in children with Down's syndrome and other aneuploid conditions. Lettieri et al 1993 conclude that in a high-risk population, an abnormally short ear length (at or below the tenth percentile) warrants further investigation to rule out a chromosomal abnormality.

We found fetal ear length to be a readily obtainable biometric measurement in the second-trimester fetus. In our study, ear length was include with routine sonographic biometry and did not appreciably prolong the examination. Our study establishs a normogram for fetal ear length by gestational age at 16-19 weeks and demonstrates that the ear length increases linearly with gestational age in the second trimester. We found that all aneuploid fetuses have ear length that are abnormally short. Our study, mean ear length of 16-19 weeks pregnancy were 9.2 mm, 10.1 mm, 10.6 mm, 11.4 mm. respectively. We found abnormal chromosome fetuses

in 4 cases, at 16 weeks 1 case was 46XX, 16P+ and ear length was 8.5 mm. 2 cases at 17 weeks were 47XY+21, 47XY+13 and ear length were 9.4 mm, 9.1 mm. 1 case at 18 weeks was 47XY+21 and ear length was 10.2 mm. Mean ear length of normal chromosome fetuses between 16-19 weeks were 10.4 mm. The prevalence of aneuploid fetuses in our study population was 0.95%.

In conclusions due to the small sample size and few abnormal chromosome detected we cannot be absolutely definite about ear length to predict abnormal chromosome fetus.

References.

1. Benacerraf BR, Gelman R, Frigoletto FD. Sonographic indentification of second-trimester fetuses with Down's syndrome . N Engl J Med 1987;317:1371-6.
2. Benacerraf BR, Miller WA, Frigoletto FD. Sonographic detection of fetuses with trisomy 13 and 18 : Accuracy and limitations. Am J Obstet Gynecol 1988; 158: 404-9.
3. Nyberg DA, Resta RG, Luthy DA, Hickok DE, Mahony BS Hirsch JH. Prenatal sonographic findings of Down syndrome : Review of 94 cases. Obstet Gynecol 1990;76:370-7.
4. Ginsberg N, Cadkin A, Pergament E, Verlensky Y. Ultrasonographic detection of the second-trimester fetus with trisomy 18 and trisomy 21. Am J Obstet Gynecol 1990;163:1186-90.
5. Adams MM, Erickson JD, Layde PM, Oakley GP. Down' s syndrome : Recent trends in the United States. JAMA 1981; 246:758-60.
6. Palomakl GE, Knight GJ, Haddow JE, et al : Prospective intervention trial of a screening protocal to identify fetal Trisomy 18 using maternal serum alphafetoprotein, unconjugated oestriol, and human chorionic gonadotropin. Prenat Diagn 1992;12:925-30.
7. Haddow JE, Palomaki GE, Knight GJ, et al. Prenatal screening for Down's syndrome with use of maternal serum markers. N Engl J Med 1992;327:588-93.
8. FitzSimmons J, Droste S, Shepard TH, et al. Long-bone growth in fetuses with Down syndrome. Am J Obstet Gynecol 1989;161:1174-7.
9. Rotmensch S, Luo J, Liberati M, et al. Fetal humeral length to detect Down syndrome. Am J. Obstet Gynecol 1992;166:1330-4.
10. Hill LM, Guzick D, Belfar RL, et al. The current role of sonography in the detection of Down syndrome. Obstet Gynecol 1989;74:620-3.
11. Nyberg DA, Resta RG, Hickok DE, et al. Femur length shortening in the detection of Down syndrome: Is prenatal screening feasible? Am J Obstet Gynecol 1990;162:1247-52..
12. Benacerraf BR, Mandell J, Estroff JA , et al. Fetal pyelectasis : A possible association with Down syndrome. Obstet Gynecol 1990;76:58-60.
13. Scioscia AL, Pretorius DH, Budorick NE, et al. Second trimester hyperechoic bowel and chromosomal abnormalities. Am J Obstet Gynecol 1992;167:889-94.
14. Benacerraf BR, Harlow BL, Frigoletto FD. Sonographic demonstration of hypoplasia of the middle phalanx of the fifth digit : A finding associated with Down syndrome. Am J Obstet Gynecol 1988;159:181-3.
15. Mith DW. Recognizable Patterns of Human Malformation. Vol VII. In the series Major Problems in Clinical Pediatrics. Philadelphia : WB saunders;1982: 11-23,30-31,72-5.
16. Birnholz , JC, Farrell EE. Fetal ear length. Pediatrics 1988;81:555-8.
17. Lettieri L, Rodis JF, Vintzileos AM, Feeney L, Ciarleglio L, Craffy A. Ear length in second - trimester aneuploid fetuses. Obstet Gynecol 1993;81:57-60.