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Brief communication

First trimester Down Syndrome screening by nuchal translucency in a Thai population

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Down syndrome (DS) is one of the most common causes of serious congenital abnormality and severe mental handicap. The incidence of DS is approximately 1 in 800–1000 [1]. The traditional method to determine the high-risk group is by maternal age-invasive testing in pregnant women aged 37 years or older, which identifies approximately 30% of fetuses with trisomy 21 [2]. The sensitivity of the DS screening test of the combined maternal age and nuchal translucency thickness (NT), which is the area between the skin and cervical spine fascia of the fetus, is approximately 80% [2].

This prospective cohort study aims to examine the significance of the fetal NT at 10–13 weeks' gestation in the prediction of DS in a Thai population. A total of 2353 pregnant women were included in the study from January 1996 to June 1999. Almost all women underwent transabdominal scanning (TAS) (Toshiba 140, TAS probe 5.0 MHz Toshiba PVF-5.00 MT). When the result showed an increased risk for DS (NT $\geq$ 2.5 mm), the woman was counseled and the invasive test was optional. All the newborns were physically examined thoroughly by pediatricians to detect the markers for major chromosomal anomalies such as DS, trisomy 18 and trisomy 13. For suspected DS or other chromosomal abnormalities, a blood sample from the newborn was sent for the chromosomal analysis.

The mean maternal age was 28.71 $\pm$ 0.13 years and 86.57% of the pregnant women were less than 35 years old. The mean gestational age was 11.94 $\pm$ 1.07 weeks. Sixty-five pregnant women had NT $\geq$ 2.5 mm and decided to have the invasive tests. Sixty had normal karyotypes while five had abnormal chromosome abnormalities (one trisomy 13, two trisomy 18 and two trisomy 21). All of

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Table 1
The sensitivity, specificity, positive and negative predictive values for trisomy 21, 18 or 13 with different cut-off values for fetal nuchal translucency thickness

NT (mm)	Sensitivity	Specificity	Positive PPV	Negative PPV
≥ 2.5	5/5 (100%)	2002/2062 (97.09%)	5/65 (7.69%)	2002/2002 (100%)
≥ 3.0	4/5 (80%)	2037/2062 (98.79%)	4/29 (13.7%)	2037/2038 (99.95%)
≥ 3.5	3/5 (60%)	2056/2062 (99.71%)	3/9 (33.33%)	2056/2058 (99.90%)
≥ 4.0	2/5 (40%)	2061/2062 (99.95%)	2/3 (66.67%)	2061/2064 (99.85%)
≥ 4.5	1/5 (20%)	2062/2062 (100%)	1/1 (100%)	2062/2066 (99.81%)

them were ≥ 35 years old. Many studies have since reported the association between increased NT and poor pregnancy outcomes as well as fetal abnormalities such as congenital heart disease, diaphragmatic hernia, and hydrops fetalis [3]. The group of 60 with normal chromosomes had detailed ultrasound scanning at 18–20 weeks' gestation and screening for maternal toxoplasmosis, cytomegalovirus, rubella virus, and herpes virus. The infection screen was negative in all cases.

There were 20 congenital heart diseases, four fetal deaths in utero at 28–30 weeks, one Bart's hydrops fetalis and 10 abortions. The rest had normal pregnancy outcomes and NT was resolved by 20 weeks. There were 316 (13.43%) patients aged ≥ 35 years. Twenty-three had NT ≥ 2.5 mm and all of them had the invasive tests which revealed five with abnormal chromosomes (as mentioned above). Two hundred pregnant women with normal NT decided to have the invasive tests due to advanced maternal age and all of them had normal karyotypes. The sensitivity, specificity, positive predictive values and negative predictive values for trisomy 21, 18 or 13 with different cut-off values for fetal NT were are shown in Table 1. This is the first report of the first trimester NT screening for DS in Thai people. The cut-off of ≥ 2.5 mm NT was used to increase sensitivity [4]. There was no abnormality detected in women aged ≤ 35 years and one case of DS would have been missed if we used the cut-off of ≥ 3.0 mm.

In conclusion, this study revealed the feasibility of NT measurement for DS screening in the first trimester of pregnancy in a Thai population in order to increase detection rate of DS in high risk pregnant women, who did not accept the invasive prenatal diagnosis procedures, as well as in the low risk group.

References

[1] Canningham FG, Gant NF, Leveno KJ, Gilstrap LC, Hauth JC, Wenstrom KD. Williams Obstetrics. 21st ed New York: McGraw-Hill, 2001, p. 944.
[2] Snijders RJM, Noble P, Sebire , Souka A, Nicolaides KH. UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation. Lancet 1998;352:343–346.
[3] Bilardo CM, Muller MA, Pajkrt E. Outcome of fetuses with increased nuchal translucency. Curr Opin Obstet Gynecol 2001;13:169–174.
[4] Snijders RJM, Pandya P, Brizot ML, Nicolaides KH. First trimester fetal nuchal translucency. In: Snijders RJM, Nicolaides KH, editors. Ultrasound markers for fetal chromosomal defects. New York: Parthenon, 1996: 121–156.