



Second Trimester Genetic Ultrasound for Down Syndrome Screening at Srinagarind Hospital

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Oral

Background and Objective: To assess the value of second trimester genetic ultrasound for screening of Down syndrome conducted at Srinagarind Hospital, Khon Kaen, Thailand.

Method: The study samples comprised of 4,033 pregnant women at high risk for fetal chromosomal abnormality, from 17th to 23th week, who had performed second trimester genetic ultrasound before genetic amniocentesis between September 1996 and December 2011. Archived medical records relating to results of genetic ultrasound and genetic amniocentesis were extracted and studied.

Result: There were 3,966 chromosomally normal pregnancies (98.3%), 43 fetuses with Down syndrome (1.1%), and 24 fetuses with other chromosomal abnormality (0.6%). Thirty of 43 (69.8%) fetuses with Down syndrome had abnormal genetic ultrasound. The

overall sensitivity of second trimester genetic ultrasound for detecting Down syndrome was 69.8% with a false-positive rate of 50.4% and likelihood ratio of 1.38. Of all the sonographic markers, short femur, and short humerus indicated the highest sensitivity at 65.1% and 44.2%. According to likelihood ratio (LR+), chest abnormality, 2 vessel umbilical cord, and facial abnormality, including cleft lip and palate, have highest LR+ of 61.49, 46.12, and 46.10, and had sensitivity at 4.7% 2.3%, and 2.3% respectively.

Conclusion: The sensitivity of second trimester genetic ultrasound for detection of fetal Down syndrome at Srinagarind hospital was rather high, and probably is an alternative method of prenatal prediction for high risk pregnant women who refused genetic amniocentesis.

Key words: Genetic ultrasound, Down syndrome screening, Second trimester

