Estimated of chromosomal abnormality rate and the maternal age-specific amniocentesis risk of trisomies 13, 18, 21 and monosomy X at Hung Vuong hospital in Viet Nam

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Objectives: To estimate of chromosomal abnormality rate and the maternal age-specific amniocentesis risk of trisomies 13, 18, 21 and monosomy X at Hung Vuong hospital in Viet Nam from October 2009 to December 2016.

Method: To collect all 623 cases chromosomal abnormalities performed amniocentesis at second trimester. Recorded 101308 cases with maternal age-specific considered as normal population who were performed nuchal translucency measurement at first trimester in Hung Vuong hospital.

Results: We detected 291 (47%) cases of trisomy 21, 29 (4.6%) cases of Trisomy 13, 99 (16%) cases of trisomy 18, 6 (0.9%) other chromosomal abnormalities, 52 (8.3%) cases of monosomy X, 61 (9.7%) % cases of sex chromosomal abnormalities, 7 (1%) cases of triploidy, 78 (12.5%) case of structural chromosomal abnormalities which were 31 cases of them were imbalance. The median of maternal age of trisomy 21, trisomy 18, trisomy 13 and monosomy X were 34, 35, 32, 30 year-old respectively, compared with normal population was 28 year-old. The risk of the fetus affected trisomy 21 with 30-year-old pregnant women, detection rates and false-positive rates were 1/413, 71%, 37%, respectively. Similarly, 35-year-old pregnant women were 1/155, 48%, 11% and 40-year-old is 1/58, 23%, 2%. The ROC curve of maternal age as a marker of trisomy 21 screening was drawn within an area under the curve (AUC) was 0.74 (95% CI 0.70 - 0.77).

Conclusion: The trisomy 21 rate was majority nearly 50% of chromosomal abnormalities. Trisomy 18 rate was the second. Maternal age of chromosomal abnormalities 13, 18, 21 was significantly higher than the normal population. The maternal age risk of trisomy 21, according to our data, was higher than the risk of normal priori risk causing the reduction of the detection rate of our Down syndrome screening program. Maternal age should not be used alone and must be combined with other markers in the Down syndrome screening such as nuchal translucency and biochemical measurements such as hCG, PAPP-A, AFP, uE3 to increase Detection Rate and decrease False Positive Rate.

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