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NON-INVASIVE SCREENING - THE PROBABILITY OF EVENTS

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Abstract- Congenital anomalies are the cause of 20.0-25.0% of cases of perinatal death, while 3.0% of children are born with malformations of varying size. We examined the predictive values and defined the credibility ratio of the combined test results. Sensitivity of the test is 94.0%, and specificity is 99.0%. The positive likelihood ratio [likelihood ratio test (LR+)] is 94.00; a negative likelihood ratio [likelihood ratio test (LR–)] is 0.06. The pretest probability that pregnant women carry a fetus with chromosomal abnormality is 1:250. Posttest odds after the combined test to discover this abnormality are 0.3760, and probability of the same case is 0.2732 if it happens that the test result is positive. The result of our study confirms the justification of combined test usage in routine clinical practice, since the posttest odds rate in the case of a positive screening increases several times over (almost 90 times); the probability of detecting a chromosomal abnormality was about 70 times.

Key words: Predictive value, combined test, ultrasonography, biochemical markers

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INTRODUCTION

Congenital anomalies cause 20.0-25.0% of perinatal deaths, while 3.0% of children are borne with malformations of varying size (Ebrahaim and al., 2000). Analytical immunochemiluminescence assays and an automated analyzer IMMULITE 2000 [Diagnostics Product Corporation (DPC), Los Angeles, California, USA] were used. Usable values of the combined test were estimated on the basis of its sensitivity, specificity and possibility of the disease in case the result was positive. By combining the values of pregnancy-associated plasma protein A (PAPP-A) and free β-subunit of choriogonadotropin (free β-HCG) in serum with nuchal translucency (NT) diameter (combined test), the possibility of detecting trisomy 21 rises up to 90.0% with 5.0% false-positive findings (Alfirevic and Neilson, 2004). The testing was done between 11 and 13^{+6} weeks of gestation. If the result was positive, some invasive methods of prenatal diagnosis were suggested to the pregnant woman. A limit value of the combined test was 1: 250. A special problem was the test result interpretation. According to the literature, 32.0% of pregnant women did not know what the term "high risk" meant after getting the results and talking to the doctor (Spencer, 2001). The research objective was: 1) to examine the sensitivity and specificity of ultrasonographic (NT) and biochemical (free β-HCG and PAPP-A) markers as parameters of the combined test, and amniocentesis in diagnostics of congenital fetal anomalies; and 2) to set the credibility ratio of the combined test results.