

Topic: Non-invasive Prenatal Testing

Title: NON INVASIVE TESTING USING IN HOUSE WHOLE GENOME APPROACH
ON ILLUMINA SMALL BENCHTOP SEQUENCER

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NIPT (non invasive prenatal testing) is highly sensitive and reliable screening test for common aneuploidies T21, T18 and T13 accompanied with neglectable risk for woman and her baby. There are approximately 50 000 pregnancies annually in Slovakia and all pregnant women undergo biochemical screening and ultrasound. Amniocentesis or CVS is offered to pregnant women 35 (38) age and above 1:250 T21 risk. Estimated annually 3500 amniocenteses (due to HR for T21) are performed, however only 1 in 34 is true positive. Last year we launched our in-house TRISOMY test (TT) which is meantime successfully implemented in clinical practise in Slovakia. During one-year period we analyzed almost 1500 samples and correctly identified 35 trisomic pregnancies. Among trisomic pregnancies 24 were T21 cases (14 XY and 9 XX, 1 monozygotic XY twins), 6 T18 cases (5 XY and 1 XX) and 5 T13 cases (3XY and 2XX). Subsequently, all of them were confirmed by invasive procedure. Compared to other screening methods, TT has a higher sensitivity and specificity of the disorders monitored, minimizes false positive results, reduces the number of necessary amniocenteses and can also determine the sex of unborn baby. We reported 1 false positive T21 case where mother was a carrier of duplication on chromosome 21 which contributed on positive signal. 6 cases did not meet criteria for assessment (0,4%) due to low fetal fraction and were not reported. Our TRISOMY test exhibits 100% sensitivity for all three common trisomies and above 99% specificity. The lowest and the highest z score value for T21 was 4,86 and 19,32, 9,54 and 27,58 for T18 and 6,11 and 30,74 for T13, respectively.