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DETERMINATION OF COMMON CHROMOSOMAL ABNORMALITIES WITH NIFTY TEST (NON-INVASIVE FETAL TRISOMY TEST) IN TURKEY

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Thanks to scientific developments, detection of many common chromosomal anomalies in the prenatal period has great importance on mother and baby health. Next-generation sequencing technology provides early detection and diagnosis in many medical situations. In this study, Nifty Test (noninvasive fetal trisomy) based on Next Generation Sequencing technology is used to determine fetal chromosomal aneuploidies in the prenatal period. NIFTY (noninvasive fetal trisomy) Test is a kind of the NIPD (noninvasive prenatal diagnosis) tests which determine common trisomy abnormalities (T21, T13, T18) along with detection of 16 different chromosomal abnormalities in fetus chromosomes from cfDNA. The test was performed by using 5-10 ml peripheral blood samples from pregnant women. Wet-lab steps of the test are formed from plasma separation, DNA isolation, library preparation, and sequencing. This part of the test was performed in Turkey while sequencing data results were analyzed and reported by BGI (Shenzhen, China). Between 2013-2018 (until April), Nifty Test was studied on approximately 15.500 pregnant women samples aged 17-57 years in Turkey. 15.250 of the Nifty Test results were negative. High risk was detected in approximately 350 patients and suggested to be directed to invasive prenatal diagnosis, thus confirming the Nifty test results. In addition to detection of fetal aneuploidies by Nifty test, maternal cancer was detected in two of the patients. The studies and research for these two cases continue in communication with BGI and patients' physicians. As a result of this study, valuable statistical data were obtained based on patients profiles and it has been indicated that preferability of NIFTY test has been increasing according to other biochemical-conventional tests and competing NIPD tests in Turkey and near countries because of its high accuracy, sensitivity and specificity ratio.

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