



Characteristics of women choosing cf-DNA testing in screening for aneuploidies

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Objective

The use of prenatal cf-DNA testing as a highly accurate screening method for aneuploidies is bringing radical changes in the field of fetal medicine. It results in decline of numbers of invasive procedures in women with high risk, based on maternal age and/or NT screening due to substitute for cf-DNA testing instead. The average age of women deciding to have cf-DNA testing is similar in the time period of last 4 years, however the numbers of tests are increasing.

Methods

We present the characteristics of pregnant woman with unselected pregnancies, on whom prenatal cf-DNA testing was performed in private outpatient clinic and public health Department of perinatology in the period from 1. 1. 2013 until 26. 4. 2017. In our study, 457 pregnant women were included. All samples were analyzed in clinical laboratory – the ISO/IEC 17025-certified clinical laboratory of BGI-Shenzhen, China.

Results

In 2016, of total 169 pregnant women with cf-DNA testing 90 (53.25 %) were of advanced maternal age (37 years or more); 39 (23.08 %) were at high risk for T21, based on prior screening test results; 27 (15.98 %) women were at high risk regarding age and prior screening test results; 67 (39.64 %) had low risk for chromosomal abnormalities regarding age and prior screening test results. The age of women with cf-DNA testing is similar in the last 4 years (2013: 36.1 years, 2014: 36.2 years, 2015: 36.4 years, 2016: 36.3 years). The number of women with cf-DNA testing is increasing over the last 4 years (2013: 39, 2014: 86, 2015: 91, 2016: 180). In general, cf-DNA testing had 100 % (95% CI: 63.1%-100%) sensitivity and 99.4 % (95% CI: 98.0%-99.9%) specificity, PPV turned out to be 80 % (95% CI: 44.4%-97.5%) for all aneuploidies. Regarding only T21, sensitivity and specificity were 100 %.

Conclusion

Our results confirmed that prenatal cf-DNA testing represents highly accurate approach for advanced screening of most common aneuploidies. The numbers of tests are increasing; however, the average age of women, opting to have cf- DNA screening, remains the same.

