Objective

To evaluate the performance of PraenaTest® for trisomy 21/13/18 in a routine clinical setting. Secondary objective was the analysis of indications to perform non-invasive prenatal testing (NIPT) and the rate of invasive testing.

Methods

A total of 2733 patients were recruited between February 2013 and July 2015 for prospective screening using PraenaTest® in Germany. Pregnancy outcome was obtained by standardized questionnaires. Complete follow-up was provided in 2194 cases so far, all being tested for trisomy 21 and n=1913 samples with analysis for trisomy 13/18.

Results

Median maternal age was 36.8 years (range 20-60), with 63.8% being > 35-years. The majority of testing was performed around 14 weeks of gestation. Advanced maternal age was stated as the most common indication for testing (47.1%). In 54.5% first-trimester screening was performed. In 0.98% (21/1913) fetal fraction was below the threshold necessary for successful analysis. PraenaTest® correctly identified all 44 cases of trisomy 21, 7 cases were tested positive for trisomy 18 and 3 cases for trisomy 13, respectively. Invasive testing was performed in 2.4% (n=52). Of those n=7 had a normal NIPT result, n=5 could not be analysed and n=40 had an abnormal NIPT result. Sensitivity and specificity for trisomy 13 and 21 was 100%, sensitivity for trisomy 18 was at 75% with a specificity of 99.8%. There were 4 false positive results and one false negative result for trisomy 18.

Discussion

NIPT is currently recommended to women with a high-risk profile for chromosomal abnormalities although there is an increasing demand from women with a low-risk profile. PraenaTest® shows a high sensitivity and specificity for trisomy 21/13/18 in a routine clinical setting. Given the possibility of false-positive test results, NIPT should always be interpreted in combination with a targeted ultrasound examination.

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