Clinical Follow-Up Study of PraenaTest®- Preliminary Results of a German Multicenter Post Market Clinical Follow-Up Study

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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

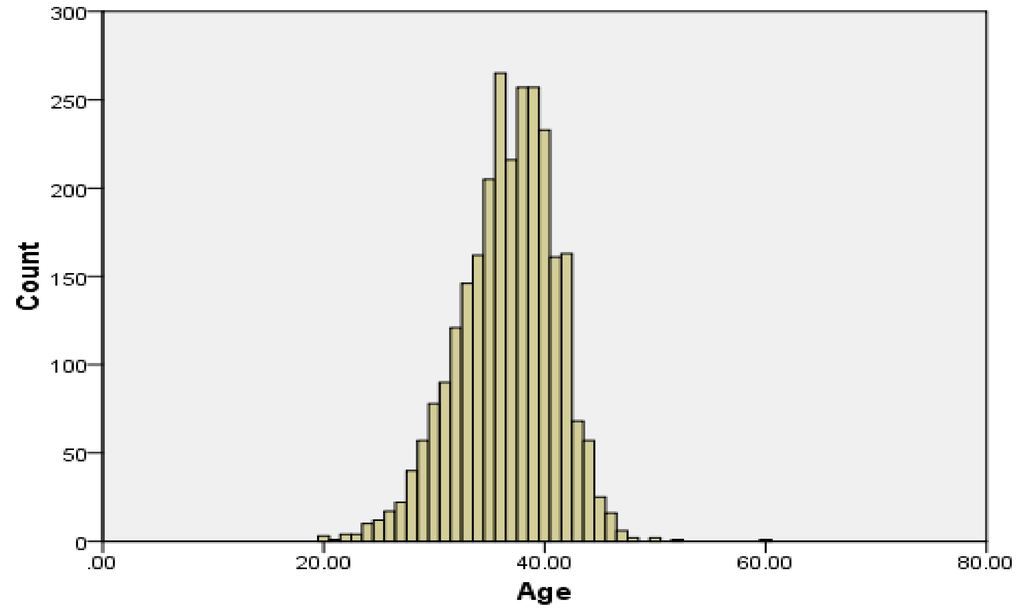
Median maternal age was 36,8 years (range 20-60), with 63,8% being > 35-years. The majority of testing was performed around 14 weeks of gestation. Advanced maternal age was stated as the most common indication for testing (47,1%). In 54,5% first-trimester screening was performed. In 0,98% (21/ 1913) 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.

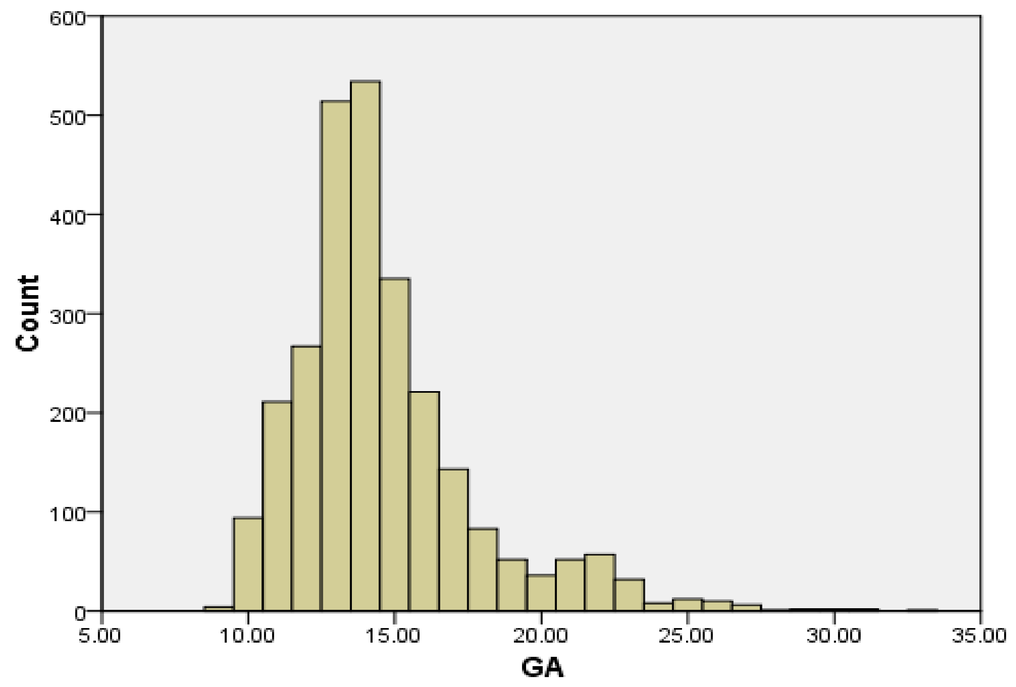
Discussion

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.

Median Maternal Age



Timing for Testing



Reasons for Testing		
Maternal Age	n=1030	47,1%
Abnormal Biochemistry	n=406	18,6%
Ultrasound Anomalies	n=402	18,4%
Maternal Anxiety	n=111	5,5%
Maternal Wish	n=132	6,2%
Previous History of Aneuploidies	n=65	3,0%
Other reasons	n=27	1,2%

Distribution of aneuploidies				
Aneuploidy	DR (%)	PPV (%)	Specificity (%)	NPV (%)
Trisomy 21	100	100	100	100
Trisomy 18	75	42,9	99,8	99,9
Trisomy 13	100	100	100	100

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