Clinical Validation of The IONA® test: A non-invasive prenatal screening test for Trisomy 21, 18 and 13
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Objective
The aim of this study was to assess the screening performance of the IONA® test for diagnosis of three fetal aneuploidies (Trisomies 21, 18 and 13) in pregnancy. The IONA® test, developed by Premaitha Health Ltd, uses an automated semiconductor sequencing platform.

Methods
This was a prospective, multicentre, blinded study. Pregnant women at increased risk of Trisomy 21 and who opted to undergo diagnostic genetic testing were invited to participate in the study, and donate a blood sample. Cell-free (cf) DNA analysis was undertaken using the IONA® test; the primary outcome of the study was the likelihood ratio and maternal age-adjusted probability result for Trisomy 21 generated by the IONA® test in the prediction of the definitive karyotype determined by amniocentesis and chorionic villus sampling (CVS) results. We also evaluated the screening performance for Trisomies 18 and 13.

Results
442 pregnant women were eligible for this study and results were available from 437 (98.9%). Samples from 5 pregnant women did not meet the validity criteria applied by the analysis software incorporated in the IONA test system (2 samples did not have sufficient DNA fragment count, 3 samples did not contain sufficient fetal DNA) and were excluded from subsequent analysis (overall sample failure rate of 1.1%). The median maternal age was 35 years and the median gestational age at enrolment to study was 15 weeks and 3 days. The typical turnaround time from start of sample processing to availability of result was 3 days. Using a risk cut-off of 1 in 150 on the age-adjusted probability result, the detection rate of the IONA® test was 100% for Trisomies 21, 18 and 13 for, with a false positive rate of 0%.

Conclusion
This study indicates that the IONA® test offers accurate non-invasive prenatal screening for Trisomy 21, 18 and 13. This can be achieved in a significantly shorter timeframe than is currently available. The rapid turnaround time would facilitate wider implementation and is likely to be a useful addition to the current screening methods for trisomy 21, 18 and 13.