Clinical laboratory experience with noninvasive prenatal testing: update on clinically relevant metrics

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Objective

The verifi® noninvasive prenatal test for high risk pregnancies has been available through our accredited clinical lab since February 2012. Many professional societies, including ISPD have published statements in support of the use of NIPT and have recommended continued monitoring of test performance for quality purposes. In follow-up to our first published clinical experience paper (Futch et al, June 2013), this study highlights our continued effort to provide clinically relevant metrics on test performance of aneuploidies for 21, 13, and 18 in over 34, 000 cases.

Methods

An active outcome follow-up process was utilized to collect pregnancy outcome information for cases with aneuploidy detected results for T21, T18 and T13. For cases with no aneuploidy detected results, clients were encouraged to report any discordant results to the laboratory.

Results

The outcome information was reviewed for a total of 34, 646 cases. Results were reported in 34, 092 cases (98. 4%) with an average turn-around-time of 3. 9 business days. Out of 554 (1. 63%) total cancellations, the majority 528 (1. 55%) were for administrative reasons; while only 23 (0. 076%) were technical cancellations. The median gestational age was 13 weeks (range 10-38 weeks) and the average maternal age was 37 years. Aneuploidy was detected in 874 (2. 56%) samples; with 596 detected T21 (1. 74%), 208 detected T18 (0. 61%) and 70 detected T13 (0. 21%). Outcome information was available in 34% of cases. There were 61 (0. 18%) putative false positive cases (18 T21, 29 T18, 14 T13) and 8 (0. 02 %) false negative cases (4 T21, 3 T18, 1 T13). Although the PPV varied with the chromosome of interest, NPV was over 99. 98% for all the three chromosomes. No significant differences were noted in US versus international samples with respect to maternal and gestational age distributions or the aneuploidy detection rates.

Conclusion

Continual monitoring of the noninvasive test performance in the clinical setting is important for assuring accurate and reliable results, as patients face decisions regarding invasive prenatal tests. The verifi® noninvasive prenatal test using increased sequence tags on an MPS platform with our proprietary algorithm, continues to show excellent test performance in the clinical setting. .