INTRODUCTION

It is well established that 50-70% of clinical miscarriages are caused by aneuploidies, mostly trisomies. To date, conventional cytogenetic and advanced molecular techniques are used for the analysis of Products of Conception (POC) to identify the genetic cause of miscarriage, providing valuable information for genetic counselling. However, both approaches are based on the direct analysis of the abortive tissue, which entails several limitations and risks including culture failure and/or maternal cell contamination (MCC), and surgical complications. Maternal cfDNA testing emerges as a promising tissue-independent alternative.

RESULTS

We found no significant differences in the NIR between iPOC and niPOC analysis [10.0% (12/120) vs. 16.7% (20/120)]. Out of 120 samples, 90 provided an informative result on fetal tissue in iPOC and niPOC groups (75%). Excluding triploidy (not tested by niPOC) cfDNA analysis correctly identified 74/87 (85.1%) samples. Sensitivity and specificity were 79.4% and 100%, respectively; all discordant cases were female.

CONCLUSIONS

Genome-wide cfDNA-based screening provides a simple, non-invasive approach to determine whether fetal aneuploidy explains the loss in early pregnancy loss or recurrent pregnancy loss (RPL) patients. Given the simplicity and accuracy of niPOC, clinicians will be able to perform genetic testing in a group of patients where it was not previously feasible.