Reviewer's report

Title: Noninvasive Fetal Trisomy Test (NIFTY) An Advanced Noninvasive Prenatal Diagnosis Methodology for Fetal Autosomal and Heterosomal Aneuploidies

Version: 3 Date: 20 March 2012

Reviewer: ec tan

Reviewer's report:

Major

1. The paper is difficult to read. Extensive editing needs to be done for grammar and language structure.

2. A number of terms which are not of common usage and/or whose meaning is ambiguous are used without defining them first: They include
   p5 para3 line9-10: heterozygous fetus
   p6 para2 line8: heterosomal aneuploidies (do they mean sex chromosome aneuploidy?)
   p8 para2 line10 and line12: tag number
   p9 para2 line3: Q20%, PCR duplication rate
   p9 para 2 line6: RIGID

3. The gestation age of the 903 samples range from 10 – 34 weeks. As it has been documented that fetal DNA concentration increases with gestation age, do the samples whose aneuploidy state which are not correctly identified have the same gestation age as those which are? In other words, for the 3 normal samples identified as trisomy 18, and the one XO case which is incorrectly identified, what gestation ages are they? For the mosaic XO case, what is the faction of normal cells?

Minor Revisions

1. In the figures, many of the labels for the X- and Y-axis are missing. In cases where the label is stated, the unit of measure is frequently missing.

2. References are not properly cited. Eg there are three papers by Lo et al in the reference list, which paper are they referring to on p10 para2 line4 as there is no reference number? And there is no Quake et al (p10 para2 line10) in the list of references.

Minor Discretionary

Instead of introducing new terms like heterosomal aneuploidy (p6 para2 line8) or heterosomal abnormalities (p9 para3 line9), the authors should just use the commonly used term “sex chromosome” aneuploidy or abnormality.
Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Not suitable for publication unless extensively edited

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:

I declare that I have no competing interests.