Validation of fetal DNA fraction estimation and its application in noninvasive prenatal testing for aneuploidy detection in multiple pregnancies (ID:1054)

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To analyze the fetal fraction, fetal sex and chromosomal aneuploidy in multiple pregnancies using noninvasive prenatal testing (NIPT).

**Objectives**

To analyze the fetal fraction, fetal sex and chromosomal aneuploidy in multiple pregnancies using noninvasive prenatal testing (NIPT).

**Methods**

A total of 362 pregnant women including 203 singleton pregnancies, 69 twins and 90 higher-order multiple pregnancies were recruited. Fetal fractions estimated by size ratio-based and Y chromosome-based approach in singleton pregnancies with male fetus were used as training data to establish the model. The model was applied to multiple pregnancies for fetal fraction estimation. By comparing the fetal fractions estimated by size-ratio to those by Y chromosome or autosomal chromosomes, fetal sex and chromosomal aneuploidy were further analyzed.

**Results**

The size ratio-based approach was well established in estimating fetal fractions for twin and higher-order multiple pregnancies. Fetal fraction had a positive correlation with gestational age in twin and triplet pregnancies. Fetal sex was correctly identified in 68 of the 69 twins and 83 of the 85 triplet pregnancies. Fetal chromosomal aneuploidies were detected in seven cases, with two singleton pregnancies and five higher-order multiple pregnancies.

**Conclusion**

Fetal sex and chromosomal aneuploidy in multiple pregnancies can be determined using NIPT.

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