Our study aimed to evaluate the performance of noninvasive prenatal testing of Trisomies 21 and 18 in twin pregnancies by maternal plasma sequencing. Pregnant women with live twin fetuses were recruited, with careful pre-test counseling, from six hospitals during April to December 2012 for this study. All MPS-based tests were performed prior to the recording of karyotyping information and the sequencing lab was blinded.

All samples performed karyotyping according such indications as follows: i) positive results in maternal serum screening tests, ii) increased nuchal translucency (NT), iii) absence of fetal nasal bone, iv) abnormal ultrasound findings in second trimester, V) twin pregnant women by IVF.

5ml peripheral venous blood sample was obtained 30 minutes before invasive procedures. Maternal plasma was isolated within eight hours by a double-centrifugation protocol and stored at -80°C. DNA was extracted from 600ul maternal plasma and sequenced on Illumina HiSeq 2000 platform. For each sample, the report was delivered within 12 days after blood sampling.

The maternal age ranged from 21 to 40 years old and the gestational age from 11th to 27th weeks. With the same pipeline for the singleton pregnancy, we correctly identified two cases with discordant fetal Trisomy 21 and one case with discordant fetal Trisomy 18. The rest 125 samples were classified as negative. Compared with the results of full karyotyping, the estimated sensitivity and specificity for trisomies 21 and 18 were 100%.

Our study suggested that NIPT for Trisomies 21 and 18 by maternal plasma DNA sequencing is of high sensitivity and specificity in twin pregnancies. It has the potential to be used as an alternative option of prenatal test for twin pregnancies.