Objectives

To assess the factors that are associated with (a) common and atypical chromosome abnormalities, and (b) the uptake of cfDNA testing vs IPD alone following positive cFTS.

Methods

A historical cohort of 1,855 Chinese women carrying singleton pregnancies with positive cFTS (≥ 1:250 for T21 or 1:180 for T18) in one public hospital over a five-year period. After counseling, women can choose cfDNA testing which was self-financed or IPD which was publicly funded. All chromosome abnormalities were confirmed by IPD with karyotyping ± aCGH.

Results

The prevalence of common and atypical abnormalities was 5.9% and 1.2% respectively.

In multivariable binary logistic regression analysis, four factors (T21 risk > 1 in 100, a high risk of both T21 and T18, increased NT, or pregnancy associated plasma A (PAPP-A) <0.2 multiple of medians (MoM)), were significant predictors of common abnormalities while and only two factors (increased NT and PAPP-A< 0.2MoM) for atypical abnormalities.

Conclusion

Our study has identified the factors associated with common and atypical abnormalities, and uptake of cfDNA testing vs IPD after positive cFTS. Low PAPP-A was a significant predictor of atypical abnormalities, but not of accepting IPD.