Non-invasive prenatal testing (NIPT) for common chromosomal aneuploidies: data from a single center in a routine screening population

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Objective
Non-invasive prenatal testing (NIPT) by analysis of cell-free DNA (cfDNA) from maternal blood has shown promise for highly accurate detection of common fetal trisomies. We assessed the performance of NIPT for common chromosomal aneuploidies screening in a routine pregnant population from a single center in Zagreb.

Methods
We present the results of prenatal cfDNA testing in a period from 25.03.2013. until 01.01.2022. in a private Hospital in Zagreb. Of total 1934 pregnant women on which cfDNA testing was performed, 19 were twin pregnancies. All samples were analyzed using massively parallel sequencing, in clinical laboratory of BGI – Shenzhen, China, and from January 2019 in laboratory Geneplanet in Zagreb.

Results
Results were available in 1930 cases (99.79%), among them delayed results in 57 (2.94%), 20 needed resampling (redraw rate 1.03 %) and only 4 ‘no call’ result (failure rate 0.20%). Among 35 (1.97%) high risk results there were 20 Trisomies 21, 2 Trisomies 18, 1 Trisomy 13, 6 SCA, 1 CriDu Chat, 1 Trisomy 5, 1 Trisomy 2 and 1 duplication 9 chr. All cases of T21, 1 case of T18 and T13 were confirmed by karyotyping (PPV for T21 is 100%). 2 women with high cf-DNA test result for T18 had miscarriage before karyotyping. Trisomy 2 was not confirmed by standard karyotyping but interestingly suspected trisomy 5 confirmed mosaic of trisomy 5 in 6% of cultured cells (it was a missed ab at 17 weeks of pregnancy). A suspected 5p deletion and was not confirmed by microarray method, but duplication of 9 chr was confirmed (mother is the carrier). There was one case of false negative T21, resulting in sensitivity for T21 of 95.2%.

Conclusion
The performance of screening for Trisomy 21, 18, 13 and sex chromosome aneuploidies by cf-DNA testing using massively parallel sequencing is most effective screening method with high detection rates and extremely high PPV (100 %) for Trisomy 21. Our results show extremely low redraw and ‘no call’ rate.