NIPT in the diagnosis of failed pregnancies

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Background: The implementation of non-invasive prenatal testing (NIPT) as a highly accurate screening test for aneuploidy is now standard practice in the field of prenatal diagnosis. One of the other possible applications of NIPT could be in the field of research on failed pregnancies. About a third of QF-PCR examinations of the products of conception cannot be performed because of high contamination of the material or for sampling of the maternal tissue exclusively. In these cases, a valid reset can be obtained accurately using non-invasive prenatal testing. We present the results of our two-year study.

Methods: A prospective study of cell- free DNA (cfDNA) testing of pregnancies with diagnosed missed abortion or miscarriage between 5 and 12 weeks was performerd using the whole-genome sequencing (Low-Pass WGS). We use the Wisecondor X (Ghent University) and Nexus copy number 10 program analyzing whole-genome copy number variants (CNVs). We estimate the fetal fraction by comparing the outcome of free methods: Defrag, SeqFF and ComboFF.

Results: 60 samples from nonviable pregnancies were analysed using QF-PCR method. The average gestational age was 8 weeks. Out of these, euploid fetuses were detected in 40 % of samples, aneuploid fetuses were detected in 22 % of samples, and in 38 % of cases it was not possible to obtain a valid reset due to maternal tissue contamination of samples. In these cases cfDNA testing was able to give a result.

Conclusion: Non-invasive testing of products of conception can be used for detection of aneuploidies in nonviable pregnancies early in the first trimester, especially in cases, where samples from RCUI cannot bereliably assessed due to maternal contamination.