

Pitfalls in the development and implementation of non-invasive prenatal testing at the regional level - Trisomy test story

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Background: Non-invasive prenatal testing is currently available as integral part of the prenatal screening in many countries. Nevertheless, there is still missing the normalization in guidelines and performance analytics in the laboratories performing different types and modifications of the test focusing on their scope, methodology used as well as analytical performance and lot of issues are tuned up based on commercial and regional instead of professional preferences.

Methods: A comparison of the laboratory parameters of two different types of NIPT tests used in one laboratory will be summarized. At the same time, on the basis of long-term follow-up, various anamnestic and clinical-diagnostic parameters of samples analyzed by Trisomy test will be summarized primarily, but not only, in the context of regional differences in the implementation of prenatal screening in three neighboring CEE countries.

Results: Based on a direct comparison of a test from the category of ready-made solutions and targeted one – Harmony, with a home-made and a whole-genome one - Trisomy test, the home-made one showed significantly lower uninformative rate (after first sample analysis). Additionally, detailed Trisomy test performance parameters were determined and represented by the PPV values of the detection of aneuploidies of chromosomes 21, 18, 13, X, Y, RATs and subchromosomal aberrations in the range of the entire genome. And at the same time differences were also identified in the regional specifics of the implementation of prenatal screening in the form of NIPT, as well as in critical parameters that are related to the different performance of the same test in cohorts of pregnant women from three neighboring CEE countries.

Conclusion: There is also lack of systematic and comprehensive comparison between the NIPT tests covering also regional specifics, although data of laboratories and countries that enabled the NIPT on population scale are now being published.