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Prospects of application and efficiency of prenatal diagnostic methods: screening for aneuploidies

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Objective

Prenatal diagnosis is crucial in the detection of hereditary and congenital diseases. It prevents births with severe and often fatal genetic and chromosomal diseases. Well-coordinated interaction of specialists in different fields involved in the prenatal diagnosis and the availability of a unified laboratory and clinical methods as well as the use of efficient screening programs with proven algorithms increase the effectiveness of the current program.

Methods

This a study that included patients in the first trimester (11w-13w+6) between in 2016 and 2017. Patients had a dating scan and a first trimester screening for chromosomal abnormlities (nuchal translucency, b-HCG and PAPP-A). The results were processed in the Astraia program, licensed by FMF, 2017 - new version. All pregnant women with a high risk for chromosomal abnormalities were referred for a genetic consultation to discuss the option of invasive testing.

Results

During the study period 4601 pregnant women were included. 148 (3. 2%) pregnant women were high risk for aneuploidies (2016 - 3. 8%; 2017 - 2. 1%). Invasive testing was performed in 82 case (33. 7%) (2016 - 61 (64%); 2017 - 21 (39%)). 64 patients declined invasive testing (43%) (2016 - 34 (35%), 2017 - 32 (60%)). 18 patients (12%) opted for NIPT (2016 - 7 (7. 3%), 2017–11 (20%)). 20 aneuploidies were confirmed (24. 3%) (in 2016 – 10 (16%), 2017 - 10 (47%)).

Conclusion

During the study period, the detection rate of chromosomal defects in the first trimester improved due to the use of the combined test with the FMF algorithm. Our data show also that the rate of invasive testing is high risk women is declining due to the fear of pregnancy loss. NIPT however is increasingly preferred as an alternative to invasive testing despite questionable results. Unfortunately this test is still expensive and hardly affordable.