Streszczenie w języku angielskim

Despite the significant progress in the diagnostic tools applied for prenatal screening, amniocentesis is still used to confirm abnormal fetus karyotype. Invasive testing carries a potential risk of miscarriage; therefore, screening biomarkers are commonly used before undergoing invasive procedures. The introduction of novel screening methods may subsequently reducing the number of incorrect indications for amniocentesis diagnosis. In the future, the introduction of integrated omics methods into routine non-invasive prenatal screening could increase the detection rate of fetal aneuploidy including T21. Based on our literature search, it can be concluded that cbDNA and cffDNA analysis demonstrate the vast potential in NIPT. However, there is still a need to provide useful data in order to validate their usefulness. Moreover, the development of fully automated systems remains essential to introduce modern technologies in prenatal screening. Accordingly, novel approaches have provided new insights into the complex pathophysiology of T21, which could be further used in novel therapeutic strategy evaluation. Up to date, ELISA was a useful tool which meet the challenge of introducing results obtained with omics-based methods into daily routine diagnostics with subsequent validation of different procedures.

Furthermore, literature data emphasize the relationship of fetal chromosomal aberrations with disturbed processes dependent on the potential of oxidative-antioxidant processes. The presence of an additional chromosome 21 disrupts a number of metabolic pathways, resulting in birth defects in the fetus. In addition, given that key genes in the oxidative stress pathway are mapped on chromosome 21, the importance of oxidative stress not only in the pathogenesis of T21, but also in prenatal diagnosis should be included. In this case, in this study, we examined the possibility of using the determination of Apolipoprotein E and selected markers of oxidative stress in prenatal screening of trisomy 21.

Concentrations of Apolipoprotein E (ApoE), concentration of DNA / RNA damage products after increased oxidative stress influence (OSDP), advanced glycation products (AGE), ischemia modified albumin (IMA), alpha 1- antitrypsin (A1AT), asprosin and vitamin D concentrations were determined in both maternal plasma and amniotic fluid in trisomy 21 (T21) and euploid pregnancies. The obtained results indicate an increased level of ApoE, OSDPs DNA / RNA and asprosin protein with a simultaneous decrease in the level of vitamin D and A1AT in the study group. Diagnostic utility in screening T21 of the above parameters based on the area under the obtained ROC curve (ROC curve) was as follows: ApoE (AUC = 0.975); asprosin (AUC = 0.965), IMA (AUC = 0.880), AGE (AUC = 0.846) and OSDP DNA /

RNA (AUC = 0.506) at the T21 screening. The obtained results indicate the potential role of the use of ApoE determination and selected markers of oxidative stress in the prenatal screening tests T21, where the greatest screening utility was demonstrated by the determination of the concentration of ApoE and asprosin protein.