Prenatal Screening in Czech Republic – history, present, future

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Background:

Today's medical advances have given us the capacity to identify many diseases before they occur and at times to apply preventative measures, so that morbidity and mortality may be avoided. Large scale of tests in pregnancy exists: gestational risk in diabetes, infection, Rhesus factor and also thyroid dysfunction. Prenatal screening of the Down's syndrome (DS) is the most common testing for aneuploidy. The management of screening in the Czech Republic is similar like in other countries in the world. The DS screening strategies started by the traditional second trimester serum biochemistry (AFP, HCG, uE3 and later also inhibin A), the combine test in the first trimester (ultrasonography markers nuchal translucency and serum biochemistry - PAPP-A and free beta hCG), or combinations of results from the both trimesters - integrated test, comes about year 2000. For a true diagnosis, the chromosomes of the foetus must be examined. From 2010 is possible to use method detection of cffDNA in mother blood. Blood sampling at the beginning of pregnancy allows the inclusion of other screening procedures such as the examination of the risk of preeclampsia using the level of PIGF. From 2024, general screening of thyroid function in pregnant women will begin with the determination of TSH.

Conclusion: Although there is no doubt that genetic testing methods will continue to expand and perhaps become cheaper, it will still take a long time before we can abandon the classic combined test, not only due to its irreplaceable position in the ultrasound diagnosis of fetal defect, but also biochemical examinations cannot be neglected, and it is essential to maintain cooperation between all interested disciplines.