

Screening for chromosomal defects

Veronika Frisova

Palacky University Olomouc, Czech Republic

There are several methods of screening for chromosomal defects. The mothers worldwide do prefer the most safe, early and accurate method of screening. Currently, the most widespread and accepted one is the first trimester screening combining maternal biochemistry with ultrasound examination of fetal nuchal translucency and eventually other ultrasound markers of Down's syndrome and other chromosomal defects. Moreover, cell-free-DNA testing methods are used as the most accurate method of screening for the most common chromosomal defects. These two methods have advantage of early screening and eventually diagnosis of chromosomal defects. Second trimester biochemistry (triple or quadruple test) is much less accurate

method of screening. Integrated test combining maternal biochemistry in the first and second trimester with fetal nuchal translucency in the first trimester has a good accuracy, however its disadvantage are late results with diagnosis in the second trimester. Genetic ultrasound in the second trimester may improve accuracy of screening for Down's syndrome, but mainly plays important role in diagnosis of other, even less frequent chromosomal defects.

This presentation gives you an overview about the methods of screening for chromosomal defects with comparison of their accuracy, advantages and disadvantages.

e: veronika.frisova@gmail.com



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