12° CONGRESS of the EUROPEAN SOCIETY of Gynecology
BARCELONA 18/21 OCTOBER 2017

NON-INVASIVE PRENATAL TESTING (NIPT) AS PRIMARY SCREENING IN THE FIRST TRIMESTER OF PREGNANCY

Bartha J (ES) [1]

Cell-free (cf) DNA testing is revolutionizing prenatal care by providing a simple, highly accurate, non-invasive screening method for fetal aneuploidy. Until now, most professional organizations have recommended that NIPT is most appropriate for high-risk patients. These recommendations have been primarily based on limited evidence regarding the utility of cfDNA testing in low- or average-risk pregnant women. However more recently it has been shown that NIPT is superior to traditional screening methods, such as the Combined test in a general screening population. Consequently more and more professional organizations are adapting their guidelines and recommending first-line screening for fetal aneuploidy by means of cfDNA testing. With improving screening technology and decreasing costs of sequencing it has also become technically possible to expand the scope of prenatal screening beyond common aneuploidy. Commercial companies already offer NIPT as a screening test for sex chromosomal aneuploidy and selected microdeletions. But as test menus expand, compromises in care can occur. Adding more conditions impacts accuracy and may result in the need for more invasive testing.

[1] University Hospital La Paz