

Role of Non-Invasive and Invasive Testing in Pregnancy in Patients with IVF and PGT

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Abstract

More than five million babies are born after Assisted Reproductive Technologies (ARTs). Nowadays ARTs are applied not only for infertile couples but also for couples with monogenic diseases and chromosomal abnormalities. Prenatal genetic testing although still not accepted as a standard procedure for couples requiring IVF has been widely used to end the transmission of genetic diseases. PGT can be applied as Prenatal Genetic Testing for Monogenic diseases (PGT-M), Prenatal Genetic Testing for Aneuploidies (PGT-A) and Prenatal Genetic Testing for Structural Rearrangements (PGT-SR). The accuracy of the PGT is still not 100% and hence couples need to be counselled about the necessity of performing further non-invasive or invasive testing in pregnancy. Both PGT and NIPT (Non Invasive Prenatal Testing) are associated with false positive and false negative results due to trophoblast-derived mosaicism. First trimester combined screening test has been the gold standard for calculation of the risk for Trisomy 21, 13 and 18 with a detection rate of 95% when nuchal translucency, nasal bone, ductus venosus and tricuspid valve blood flow are assessed. It involves also early assessment of fetal anatomy and detection of major fetal defects in the first trimester. The management plan for IVF patient with PGT should be first trimester screening test, followed by comprehensive counselling and reassurance or recommendation for NIPT or invasive testing depending on the findings. The role of the IVF specialist is to recommend the correct test for the correct patient. Fetal medicine specialist should perform the necessary screening tests and advise additional testing if required or further reassurance.

Biograph :

Dr. Desislava Markova graduated from the Higher Medical University Bulgaria qualifying as a Consultant in Obstetrics and Gynecology. She was a Senior Assistant Professor in Obstetrics and Gynecology at Higher Medical University Bulgaria as well. Dr. Desislava successfully completed a three-year training and practicing of Fetal Medicine at Kings' College Hospital-London under the direction of Professor Kypros Nicolaides and was awarded the Fetal Medicine

Foundation Diploma. She also completed the Advanced Training Skills Module in Fetal Medicine at King's College Hospital-London and a master's degree in prenatal genetics and fetal medicine in University College London, UK. She has extensive experience in ultrasound scanning during the first, second and third trimesters of pregnancy and intrauterine procedures. She is skilled in diagnosing an early fetal anomaly or any chromosomal abnormality and suggests an appropriate medical plan to solve the problem out as early as possible.