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Case report: Confirming the role of conventional combined biochemical prenatal screening in first trimester for detection of genetically affected fetuses

Elena Gjorgievska Nikolovska, Aleksandra Atanasova Boshku, Iskra Dukova

University Clinic for obstetrics and gynecology, Macedonia

First trimester screening is combined test with two phases: first is ultrasound exam of the fetus, with measuring his crown ramp length and nuchal fold (the size of the clear space in the tissue at the back of the fetuses neck and second faze - blood test for measuring two pregnancy associated substances like pregnancy-associated plasma protein-A (PAPP-A) and human chorionic gonadotropin (HCG). .

This screening is giving early information about baby's risk of certain chromosomal conditions, specifically, Down syndrome (trisomy 21) and extra sequences of chromosome 18 (trisomy 18) and Trisomy 13. It is performed between 11 and 14th gestational week, which gives enough time for performing further diagnostic tests, management of the pregnancy and medical treatment.

We are presenting the case of 29 years old, primigravida, with regular menstrual cycle of 28-30 days, with duration of 5-6 days. First ultrasound exam was performed in 6th gestational week, when gestational sac was with regular form and the fetus was visible with positive hearth rate. Next examination was performed in 10th gestational week, with CRL which wasn't in the adequate standard measurement for the given week of gestation. Next examination was after two weeks, when the CRL was with three weeks growth restriction, and was indicated first trimester prenatal risk evaluation, using PRISCA software 5.0.3.1. With result for risk for Trisomy 13/18 with nuchal translucency >1:50, both biochemical parameters PAPP-A and free BHCG were low, and there adequate Corr. MoM were lower than 0,15. This indicated further genetic testing, which the patient decided to be chorionic villus sampling. The result from the genetic testing was Abnormal karyotype of a male fetus with 69 chromosomes, with additional pair of chromosomes (23), resulting in triploidy. The final decision for this pregnancy was terminating with standard procedure of surgical abortion.

Conclusion: Most countries worldwide support non-invasive double examination as ultrasound and biochemical serum test for detection of chromosomal abnormalities with accuracy of 97-98 %. Cell free DNA test have been introduced lately for detection of chromosomal abnormalities with accuracy of 99%, but with 100 folds higher price, which is unaffordable for everyone, and also high price of this test makes him unsittable for mass screening. Our case report shows that even rare cases of genetically affected fetuses can be adequately detected with conventional combined biochemical prenatal screening, and their relatively low price makes this type of screening suitable for mass population screening.

Biography

Elena Gjorgievska Nikolovska obstetrics and gynecology specialist at University Clinic for gynecology and obstetrics

nikolovska_eli@yahoo.com