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Comparative Evaluation Between First and Second Trimester Aneuploidy Screening Tests with Amnio Synthesis Results

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ABSTRACT

Background & Objective: Screening tests for common aneuploidy, especially trisomies 21, 18, and 13, should be offered to pregnant people of all ages at first and second trimesters. Theses include combined tests (nuchal translucency (NT)+biochemical pregnancy-associated plasma protein-A (PAPP-A), free beta-human chorionic gonadotropin (β-hCG)) and quad test. If the tests are positive based on cut-off value of 1:250, diagnostic tests such as amniocentesis and chorionic villus sampling (CVS) are suggested. The current study aimed to comparatively evaluate the results of these screening tests and amniocentesis.

Materials & Methods: This practical-basic study was designed to evaluate the accuracy of first and second trimester screening tests with amniocentesis results. A total of 45 subjects were selected from the patients referred for abnormal screening tests toShohadaya-ye-Tajrish hospital in Tehran, Iran, during 2014-2015. The results of tests (combined or quad) were positive based on cut-off value of 1:250. Data was collected through a questionnaire containing such information as age, gravidity, previous aneuploidy births, placenta location, blood group, amniotic fluid color, type of sampling needle, and mode of needle entrance.

Results: The mean age of patients was 33.16 years. There was significant p-value between positive results of amniocentesis and age of the patients. Out of 45 amniocentesis tests, four had abnormal results (two in the first trimester (2/17) and two in the second trimester (2/24). Also, three items were T21 and the forth was translocation between chromosome 11-22. We had one fetal loss due to amniocentesis procedure.

Conclusion: Diagnostic tests are suggested for better evaluation of abnormal results. These screening tests have false positive and negative results. We attempted to evaluate the real results with invasive tests. Further research is needed to investigate the accuracy of screening aneuploidy tests.

Keywords: Amniocentesis, Second trimester screening, First screening, Aneuploidy

Introduction

The birth of a neonate with chromosomal abnormalities (for example Down syndrome) can cause serious problems for the neonate, family, and society. Hence, it is necessary to perform prenatal screening tests to discover the most common chromosomal anomalies **(1,2).** Currently, aneuploidy screening is suggested for all pregnant women **(1)**. In this regard, first and second trimester screening tests are relatively cheap and non-invasive methods **(3)**. Invasive diagnostic tests such as amniocentesis and chorionic villus sampling (CVS) are expensive, and they have the risk of fetal loss. CVS is usually done between 11w-13w+6 days and amniocentesis between 15-20 weeks of gestational age **(1)**.

Jeans Gekas *et al.* (2014) showed that first and second trimester screening tests and amniocentesis are some cost-effective methods, and the best options to confirm neonatal abnormalities **(4)**. In a meta-analysis, Ojilvie, j BETA, R. AKOLEKAR (2015) evaluated 110 articles with 42,716 cases under amniocentesis and reported that there were 324 (0.81%) fetal losses **(5)**.

In a retrospective descriptive study, Kovoosi *et al.* (2015) showed that the incidence of aneuploidy was one in 491 births in Tehran, Iran and its prevalence increased with increasing maternal age **(6)**. Moreover, Tara *et al.* (2016) reported that the rate of miscarriage following amniocentesis was 1% in Mashhad, Iran. They also concluded that there was no correlation

between the causes of amniocentesis and mother's blood group, maternal underlying diseases, and miscarriage following amniocentesis **(7)**.

Materials and Methods

This prospective cohort study was performed on IVF caThis study compared the results of first and second trimester laboratory tests (FT1 and ST2) and amniocentesis results in 45 pregnant women (mean gestational age=16 weeks and 3 days) with positive FT1 or ST2 referred to Shohadaya-ye-Tajrish Hospital in Tehran, Iran during 2014-2015. Using a questionnaire, the following information was collected: age, abnormal screening results, gravidity, previous aneuploidy baby, placental location, maternal blood group, color of amniotic fluid (AF), type of needle, and method of needle entrance. Patient satisfaction was obtained by an informed consent letter or phone calls. The results of amniocentesis and outcome of pregnancies were followed up by phone. The data was analyzed by IBM SPSS Statistics for Windows, version 19.0 (IBM Corp., Armonk, N.Y., USA).

Results

The average age of participants was 33.16 (standard deviation=5.4; minimum and maximum age=22 and 44 years). Among the participants, 17 and 24 subjects had positive results for FT1 and ST2, respectively; furthermore, the results of both tests in four subjects were positive. Also, 44.4% of participants were older than 35 years and one patient had a previous history of Down syndrome birth. The average gestational age when performing amniocentesis was 16 weeks and 3 days. In the study group, one patient was high-risk for T18, four samples were abnormal, three had a history of Down syndrome birth, and one item was translocation 11-22 chromosome. We had one fetal loss due to amniocentesis. There was no correlation between the outcome of diagnostic tests and gravidity, previous history of Down syndrome birth, blood group, and color of AF. In correlation with placenta location and fetal loss there is no study. In our study, out of two abortions, one case was posterior placenta and the other one was posterolateral. The mean age for abnormal amniocentesis results was 33.75 years (standard deviation=5.45 years). Therefore, there was a significant correlation between abnormal results of amniocentesis and older age (P<0.05). The study was approved by the Research Ethics Committee of Shahid Beheshti University of Medical Sciences, Iran (code: 336).

Discussion

Performing amniocentesis is necessary when screening results are positive. Niromanesh *et al.*, nin a study about pregnancy outcome in amniocentesis and CVS; ten years report, between amniocentesis and outcome of pregnancy there is definite correlation **(9)**. The sensitivity of amniocentesis for diagnosing chromosomal disorders was 100% (8,9). In addition, stress management of pregnant women with positive screening tests is very important to improve pregnancy outcome **(10)**. Shirazi *et al.* (2015) compared the complications, indications, and results of screening methods with amniocentesis and CVS, and concluded that both methods are quite safe, logical, and cost-effective **(11)**. Another study carried out in 2019 showed that these diagnostic tests do not increase the adverse pregnancy outcomes such as gestational diabetes mellitus and preeclampsia **(12)**. Maternal stress because of positive aneuploidy Screening Serum Analytes may affect on color Doppler ultrasound and pregnancy outcome; so, diagnostic tests are obligatory **(13)**. Specially in misdiagnosis in ultrasound evaluation and limitations of screening tests. It seems that diagnostic tests should be offered to all pregnant women with positive screening tests. The main limitation of this study is its small sample size. Since second trimester screening tests such as prenatal cell-free DNA screening tests are expensive, and even unavailable, in Iran, amniocentesis and CVS are the best options for positive screening tests. It is recommended that new studies be conducted on pregnancy outcome in positive screening tests that their diagnostic test are negative also pregnancy loss in these groups in a larger sample size.

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Conflict of Interest

Authors declared no conflict of interests.

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