AROG Advanced Research in Obstetrics and Gynecology

OPEN ACCESS
arogjournal.org

ORIGINAL RESEARCH ARTICLE 2023, 1(1), Article ID: e2301

The Efficiency of First Trimester Screening Test: A Retrospective Study in a Referral Center in Turkey

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ABSTRACT

Objective: To investigate the effectivity of first trimester screening test applied in a referral center in Ankara, Turkey.

Materials and Methods: The records of 2015 patients who underwent the first trimester screening test between January 2016 and December 2017 were retrospectively screened. We did not take the patients who does not postpartum newborn examination, whose pregnancies resulted as abortion and who did not sustain the routine pregnancy control visits. Invasive diagnostic test was recommended to patients who has high-risk test results. Amniocentesis was applied to the patients who accepted the procedure. We reached the newborn examination results of the patients who did not have amniocentesis result. Amniocentesis was recorded with the patient's new registrar record. Sensitivity, specificity, positive predictive value and negative predictive value were calculated according to biochemical and combined risks respectively.

Results: According to the combined risk of first trimester screening test, the sensitivity was 80%, specificity was 95.9%, positive predictive value was 6,25% and negative predictive value was 99,9%. Sensitivity was determined as 60%, specificity 88.6%, positive predictive value 1.7% and negative predictive value 99.9% according to biochemical risk.

Conclusions: The first trimester screening test is an effective prenatal test for the detection of chromosomal anomalies. Adding NT measurement to biochemical parameters increases the efficiency of the test significantly.

KEYWORDS

First trimester screening test; sensitivity; chromosomal anomalies; Down Syndrome.

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How to cite this article
Öcal FD, Soysal Ç, Özdemir O, Yılmaz E. The Efficiency of First Trimester Screening Test: A Retrospective Study in a Referral Center in Turkey. *Adv Res Obstet Gynaecol*. 2023;1(1):e2301.

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Introduction

Down syndrome (trisomy 21) is the most detected trisomy among live births with an incidence of 1.3/1000.[1] Down syndrome, which is the most common cause of severe mental retardation all over the world, causes lifelong mental and social development retardation, and imposes material and moral responsibilities for both the family and the society. To diagnose this pathology during the early period of pregnancy enables to family to decision of the course of the pregnancy. Many methods have been developed for detect Down syndrome and other chromosomal abnormalities during the early period of the pregnancy. Nicolaides defined the relationship between nuchal translucency (NT) and trisomy in nineties years.[2,3]

First trimester screening test is applied in 11-14 weeks of pregnancy. First trimester screening test depending on combination of fetal NT, maternal age and maternal serum biomarker levels (pregnancy associated plasma protein-A and beta human chorionic gonadotropin) can predict 90% of fetuses with trisomy with a 5% false positive predictive ratio[4].

The aim of this study was to determine the efficiency of first trimester screening test performed in Dr. Sami Ulus Education and Research Hospital in Ankara, Turkey.

Material and Methods

The records of 2015 patients who underwent the first trimester screening test in Dr. Sami Ulus Education and Research Hospital between January 1, 2016 and December 31, 2017 were retrospectively reviewed. Among the patients who did not undergo amniocentesis, those whose results regarding the newborn could not be obtained after delivery, those whose pregnancy resulted in abortion and those who were out of follow-up were not included in the study. All NT measurements were made by two experienced radiologists with Fetal Medicine Foundation certification, using a B-mode ultrasonography device (ATL HDI 5000) and a 3-7 MHz linear probe. While the combined risk was defined as the risk obtained by evaluating the NT measurement and biochemical markers together, the biochemical risk was evaluated as the risk calculated only according to the biochemical markers. Patients with values of 1/300 and above for the first trimester combined and/or biochemical risk were divided into two groups as high-risk and those below low-risk. Invasive diagnostic tests (chorionic villus sampling or amniocentesis) were offered to all patients in the high-risk group. The obstetric results of the patients in this group who did not accept the diagnostic test were determined according to the postpartum newborn evaluation results.

According to the data obtained, the sensitivity, specificity, positive and negative predictive values of the first trimester screening test were calculated.

Results

The demographic characteristics of the patients was shown in Table 1. Of the 2015 patients who underwent the first trimester screening test, 561 were excluded from the study due to the inability to access the newborn records, the pregnancy resulting in abortion, and the patient being out of follow-up. Out of the remaining 1454 patients, the results of 64 patients (4.4%) were higher according to the combined risk, and the results of 170 patients (11.7%) were higher according to the biochemical risk.

Amniocentesis was performed for diagnosis in 30 of the patients with high combined risk. In 4 of these patients, chromosomal anomalies (1 trisomy 18, 1 monosomy 18 and 2 trisomy 21) were detected as a result of amniocentesis. Postpartum trisomy 21 was found in one of the patients with normal combined risk. Amniocentesis was performed in 26 patients in the group with high

biochemical risk. According to the results of amniocentesis, 3 chromosomal anomalies were detected (1 trisomy 18 and 2 trisomy 21). Accordingly, the sensitivity of the first trimester screening test according to the combined risk was 80%, the specificity was 95.9%, the positive predictive value was 6.25%, and the negative predictive value was 99.9%. According to biochemical risk, sensitivity was 60%, specificity was 88.6%, positive predictive value was 1.7%, and negative predictive value was 99.9% (Table 2).

Table 1. Demographic characteristics of patients

	Age	Gravidity	Parity	Gestational week
Mean±SD	22.1±2,30	2,4±1,40	1,01±1,02	12,4±0,46
Min-Max	18-26	1-11	0-7	11-14

SD: Standard deviation, Min: Minimum, Max: Maximum.

Table 2. The Sensitivity, Specificity, Positive and Negative Predictive Value of Combined and Biochemical Methods

	Combined risk (%)	Biochemical risk (%)
Sensitivity	80	60
Spesificity	95,9	88,6
Pozitive predictive value	6,25	1,7
Negative predictive value	99,9	99,9

Discussion

The first trimester screening test is a two-step test performed between 11-14 weeks of gestation. Risk calculations were made for Down syndrome and Edward syndrome by combining pregnancy-related plasma protein (PAPP-A) and human chorionic gonadotropin, -human chorionic gonadotropin (HCG)- measurement in maternal blood and ultrasonographic nuchal translucency (NT) measurement.

Biochemical risk was calculated according to the biochemical parameters measured from maternal blood in the test results, and the combined risk was calculated as a result of the combination of these parameters with NT. In the literature, the sensitivity of NT measurement alone in detecting Down syndrome was 71.43% with a false positive rate of 4.14%, while this rate increased to 86% with a 4% false positive rate when used with biochemical parameters (combined test).[5,6] Similarly, Down syndrome detection rate of biochemical markers alone has been reported as 70%, and false positivity rate as 5%.[7,8]

Numerous studies have been carried out to determine the sensitivity and specificity of prenatal screening methods in Turkey and in the World. One of the most important studies among these is the Serum Urine and Ultrasound Screening Study (SURUSS)[9]. In this study, the false positivity rate was reported as 20%, based on 85% sensitivity for NT measurement alone. In our study, the sensitivity according to biochemical parameters alone was 60% with a false positive rate of 11.4%, while this rate increased to 80% with a false positive rate of 4.1% when NT was added.

Bahadirli et al. also found that combined risk was more successful in detecting chromosomal abnormalities than biochemical parameters alone.[10]

Adding the NT value to the biochemical parameters greatly increases the sensitivity of the first trimester screening test. NT measurement, which significantly affects the test result, is valuable when performed by trained experts. Although combined risk measurement is the most useful method for screening for chromosomal anomaly risk in the first trimester, accurate and standardized measurement of NT becomes an important problem affecting the results. In addition, all parameters examined in the first trimester screening test are affected by the gestational week. For this reason, MOM (multiple of median) values of the parameters affecting the test according to the gestational week should be used and these median values should be calculated for each laboratory according to its own conditions.

According to our results, the sensitivity and specificity of the combined test was found to be similar to the values in the literature. While the number of patients with high risk according to biochemical parameters is 170, this number is 64 in combined risk. Therefore, adding NT to biochemical parameters also reduces the rate of unnecessary invasive procedures.

In conclusion, the first trimester screening test is an effective method for chromosomal anomaly screening and the combined method is superior to biochemical marker screening. Adding NT to biochemical markers also reduces the rate of unnecessary invasive interventions. However, it is important that the NT measurement is done correctly.

Conflict of interest

The authors declare that they have no conflicts of interest.

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