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Original Article

Reasons for declining prenatal diagnostic tests and analysis of factors impacting the decision

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ABSTRACT

Objective: To identify the factors impacting the decision of declining to have prenatal diagnostic tests and to determine the reasons for declining in cases with high trisomy risk in prenatal screening tests and those who refuse to undergo prenatal diagnostic tests.

Materials and methods: Cases with positive test results in first and second trimester aneuploidy screening tests were included in the study. The patients were divided into two groups those who accepted prenatal diagnostic tests (Group 1) and those who refused prenatal diagnostic tests (Group 2). The patients that refused prenatal diagnostic tests were evaluated with a questionnaire to determine the reasons for refusal.

Results: 204 cases were included in the study. 44 (21.6%) of these cases wanted to have prenatal diagnostic tests, while 160 (78.4%) refused to have prenatal diagnosis tests. A significant correlation was found between the decision to refuse the prenatal diagnostic test and the mother's educational status level (p=0.035), whereas no significant correlation was found between the personalized risk ratios and the number of abortions in previous pregnancies. Among the reasons for refusal of prenatal diagnosis test, the main reason was determined to be "Termination of pregnancy is not an option".

Conclusion: Pregnant women should be offered all options, including not having a prenatal screening test or a prenatal diagnostic test, by providing accurate and understandable information about the purpose and potential outcomes of prenatal screening tests. Hence, pregnant women can be provided with the opportunity to make an informed choice.

Keywords: decision-making; prenatal diagnosis; prenatal screening

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Introduction

Prenatal genetic screening is used to evaluate whether the fetus is at elevated risk of being affected by a genetic disorder. Initially, prenatal genetic testing focused mainly on trisomy 21 (Down's syndrome), but nowadays it can detect a diverse range of genetic disorders [1].

Various concepts have been developed for prenatal genetic screening. Evaluation of maternal serum levels of specific biochemical markers associated with Down syndrome, with or without specific ultrasound markers, is the most widely used method for prenatal genetic screening [2]. The personalized risk for fetal aneuploidies is calculated by assessing the patient's age, medical history, and the results of prenatal genetic screening tests. The screening means a positive test result when this risk exceeds a specific threshold value.

A secondary screening or diagnostic procedure is recommended for patients with a positive serum analyte screening test. The use of cell-free DNA (cfDNA) screening as secondary screening is an option for patients trying to avoid diagnostic testing [3]. The main purpose of prenatal diagnostic tests is to provide a prenatal diagnosis of fetal aneuploidies and to provide informed choices to the family as to whether they want to continue the pregnancy or not.

The aim of this study is to identify the factors impacting the decision of declining and to determine the reasons for the decline in patients with high trisomy risk in prenatal screening tests and those who refuse to undergo prenatal diagnostic tests.

Material and methods

This study was conducted in the gynecology and obstetrics clinic of a tertiary referral center between February 01st, 2021, and August 01st, 2021. The local ethics and research committee approval was obtained for the study (registration number:2021/1268). Participants were informed the purpose of the research. The patients with positive first-trimester combined test, Second-trimester triple test, and Second-trimester quadruple test results were included in the study.

In prenatal screening tests, patients with a calculated personal risk of >1:300 for Trisomy 21 and patients with a calculated personal risk of >1:150 for trisomy 13/18 were considered to be positive screening test results [4]. All patients with positive screening test results were evaluated by ultrasonography.

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Prenatal diagnostic tests or cfDNA as secondary screening were offered as an option for patients with positive screening test results. The patients that chose cfDNA screening or who had major anomalies in the ultrasonographic examination were excluded from the study. All patients were informed about the performance, complications, and adverse outcomes of prenatal diagnostic tests.

Age, gravidity, parity, history of abortion, having a baby with an anomaly, gestational week, calculated personal risks in prenatal screening tests, and educational status of all patients were noted down.

The patients were divided into two groups those who accepted prenatal diagnostic tests (Group 1) and those who refused prenatal diagnostic tests (Group 2). Patients that refused to have prenatal diagnostic tests were asked to mark the appropriate answer, given as reasons for refusal, from the questionnaire.

Data were analyzed by using SPSS version 23.0 (IBM, Armonk, NY, USA). Descriptive statistics were performed as the mean ± standard deviation. Association between declining amniocentesis and explanatory variables was verified with the chi test. p-value < .05 was accepted to be statistically significant.

Results

Throughout the study period, 204 patients were included in the study. 44 (21.6%) of these patients wanted to have prenatal diagnostic tests, whereas 160 (78.4%) patients refused to have prenatal diagnosis tests. Demographic and clinical characteristics of patients who accepted or did not accept prenatal diagnostic tests are presented in Table 1. There was no significant difference between groups 1 and 2 in terms of basic clinical features.

Table 1. Comparison of Group 1 and Group 2 demographic and clinical characteristics

Variable	Group 1 (n:44)		Group 2 (n:160)		P value
	Mean (Min-Max)	SD	Mean (Min-Max)	SD	
Maternal age	33.3 (19-49)	6.86	33.2 (18-49)	6.78	0.94
Gravidity	3.75 (1-9)	1.89	3.70 (1-11)	2.08	0.88
Parity	2.34 (0-8)	1.64	2.11 (0-10)	1.75	0.42
Abortion	0.45 (0-2)	0.76	0.58 (0-5)	1.03	0.45
Gestational age	17.3 (12-25)	2.81	17.6 (12-26)	3.25	0.57

When both groups were compared in terms of having a baby with an anomaly, a history of a baby with an anomaly was found in 3 (6.8%) patients in Group 1 and in 5 (3.1%) patients in Group 2. No significant difference was found between the two groups in terms of having a baby with anomaly (p:0.37).

The type of screening test underwent by the patients, the type of Trisomy found to be risky, the personalized risk ratios, the number of abortions in their previous pregnancies, and their educational status are shown in Table 2.

Table 2. Comparison of Group 1 and Group 2 in terms of screening test type, educational status, personalized risk score, and the number of miscarriages in the previous pregnancy.

	% n :204	Accepting n:44 (%)	Declining n:160 (%)	P value
Screening Test				0.95
First-trimester combined test	103 (%50.5)	22	81	
Second-trimester triple test	76 (%37.2)	16	60	
Second-trimester quadruple test	25 (%12.3)	6	19	
Trisomy type				0.90
Trisomy 21	194 (%95.1)	42	152	
Trisomy 13/18	10 (%4.9)	2	8	
Educational level				0.03
Literate	40 (%19,6)	7 (%17.5)	33 (%82.5)	
Elementary school	96 (%47,1)	15 (%15.6)	81 (%84.4)	
High school	40 (%19,6)	15 (%37.5)	25 (%62.5)	
University	28 (%13,7)	7 (%25)	21 (%75)	
The personalized risk scores				0.85
1 to 100	82 (%40.2)	19	82	
101 to 200	60 (%29.4)	13	47	
200 to 300	62 (%30.4)	12	50	
The number of previous miscarriages				0.71
0	139 (%68.1)	31	108	
1	36 (%17.6)	6	30	
≥2	29 (%14.2)	7	22	
A history of a baby with an anomaly		3 (%6.8)	5 (% 3.1)	0.37

A significant correlation was found between the decision to decline to have the prenatal diagnostic test and the educational status of the mother (chi^2 = 8.592, df = 3, p =0.035), whereas no significant difference was found between the decision to refuse to have the prenatal diagnostic test and the personalized risk score (chi^2 = 0.304, df = 2, p =0.859).

The most common maternal serum screening test underwent by the patients included in the study was the first-trimester combined test with 50.5%. No significant difference was found between the type of maternal serum screening test underwent and the decision to refuse to have the prenatal diagnosis test.

When the reason for refusal was analyzed in patients who refused to have prenatal diagnosis tests, the most common reason was "Termination of pregnancy is not an option".

The reasons for the refusal of the patients who decided to refuse prenatal diagnostic tests are presented in Table 3.

Table 3. Reasons for refusing to have the prenatal diagnostic test

Reasons for refuse	N , %
Termination of pregnancy is not an option	124 (%77.5)
Possible complications of prenatal diagnostic tests on pregnancy (fetal loss, rupture of membranes, direct fetal injury, indirect fetal injury, infection)	16 (%10)
I don't think my screening test is high risk	10 (%6.25)
Distrust in the results of screening tests	8 (%5)
Test too painful or too uncomfortable	2 (%1.25)

Discussion

The ultimate goal of maternal serum screening tests undergone during pregnancy is to identify patients at risk for the most common types of trisomy and to detect fetuses with trisomy by applying diagnostic tests to these selected patients. In our study, 21.6% of the patients found to be risky as a result of maternal serum screening tests accepted having a prenatal diagnosis test, whereas 78.4% refused to have a prenatal diagnostic test.

In the literature, the rate of refusal of the prenatal diagnosis test in patients with positive screening test results varies between 21 and 45% [5,6]. The 78.4% found in our study is significantly higher than the literature.

When the reasons for refusal of having prenatal diagnostic tests were analyzed in our study, it was determined that the most common reason for refusal was "Termination of pregnancy is not an option". This reason accounted for roughly three-quarters of the patients declining to have the prenatal diagnostic test. In studies investigating attitudes towards termination of pregnancy in patients where the chromosomal anomaly was detected in prenatal diagnostic tests, it was found that women who were considering termination of pregnancy in case of the chromosomal anomaly were found to prefer prenatal diagnostic tests more frequently [7-9]. Likewise, in our study, we found that attitudes towards pregnancy termination (continuing pregnancy even if the fetus is found to have chromosomal anomaly) are the main factors in accepting or refusing to have prenatal diagnostic tests.

We determined that the second most common reason for the refusal in patients who declined to have prenatal diagnostic tests were not being exposed to possible complications of prenatal diagnostic tests during pregnancy. These pregnant women stated that they would request pregnancy termination if trisomy was detected in the fetus. A second screening with cfDNA may be a reasonable option for these patients. Because the most important advantage of cfDNA over invasive tests is that it does not pose a risk of fetal loss. However, given the high cost of cfDNA screening, this option may not be reasonable for all pregnant women.

In a study analyzing the reasons for accepting or refusing to have prenatal diagnostic tests, the most common reasons for refusing to have prenatal diagnostic tests have been found to be "termination of pregnancy is not an option" and "because invasive testing increases the risk of miscarriage" [10]. The results of this study are similar to the results of our study. When other reasons for refusal of prenatal diagnostic tests are examined; Factors such as the attitudes of the pregnant woman, the pregnant woman's husband, and the physician towards prenatal diagnosis and termination of pregnancy, the mother's anxiety state, her experiences with people with Down syndrome, socioeconomic factors, and ultrasound findings during pregnancy were determined [11-13]. Although some studies found a significant correlation between advanced maternal age [14], personalized risk score [15] as a result of maternal serum screening tests, the number of previous miscarriages [16], and refusal of having the prenatal diagnostic test, no significant correlation was found in our study.

In our study, when the patients who accepted or refused to have prenatal diagnostic tests were evaluated in terms of their educational status, a significant difference was found. In our study, refusal of having prenatal diagnostic tests was found mostly at the level of literate and elementary school education. Similarly, in a study evaluating patients with the positive screening test and recommended amniocentesis, the decision to refuse amniocentesis was found to be associated with the educational status of the mother [15]. Likewise, in our study, refusal of amniocentesis was observed most frequently in patients with primary education levels.

Prenatal screening tests for pregnant women are mostly performed by obstetricians.

The situations described below may be the reasons for performing prenatal screening tests in our country without adequate information:

1- Physicians cannot spare enough time to inform the pregnant woman due to their workload

2- The physician's desire to protect himself against malpractice cases in the presence of a trisomic fetus compatible with life such as Down syndrome (not being a party to a possible malpractice case by referring him to the appropriate center if the prenatal screening tests are performed and if the screening is positive)

"Termination of pregnancy is not an option", which is the most common reason for refusal of prenatal diagnostic tests, indicates that these patients are not adequately informed during a prenatal screening test, whether they are caused by religious concerns, ethical concerns, or any other situation. Because of the nature of prenatal screening tests, the primary goal of these tests is to identify risky fetuses and direct them to prenatal diagnostic tests and identify trisomic fetuses. Hence, it is to help the family to make a conscious choice in terms of the continuation or termination of pregnancy. If the pregnant women are informed about the purpose of the screening tests and the options that will be offered when the screening test is positive, some of the patients will not accept undergoing prenatal screening tests. Thus, it will be possible to prevent unnecessary costs and loss of workforce for both the pregnant and the public.

Healthcare providers should ensure that pregnant women understand the purpose and potential consequences of prenatal screening tests by providing all options, including not having any prenatal screening or diagnostic testing. Thus, pregnant women can make more conscious decisions to refuse or accept prenatal screening or diagnostic tests. As a result of decisions made with this awareness, in pregnant women who state that prenatal screening tests will not have an impact on pregnancy follow-up and course, these tests can be avoided, and health-care costs financed by the public in most countries can be reduced.

Disclosure

Authors have no potential conflicts of interest to disclose.

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