Demographic and Clinical Profile of Women Attending Genetic Clinic for Prenatal Invasive Testing at a Tertiary Referral Center, India

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ABSTRACT

Background

The Prenatal Invasive Tests (PIT) are used during pregnancy for the detection of genetic anomalies. Studies addressing the profile of women who come to seek prenatal invasive testing are scarce.

Objective

To assess the socio-demographic characteristics and clinical profile of women who are referred for prenatal invasive testing in a tertiary referral center in India.

Method

A descriptive, cross-sectional study was undertaken in 60 women who came to the genetic clinic at a tertiary referral center, New Delhi, India following counseling regarding prenatal invasive testing. Data was collected using a self-developed and validated semi-structured questionnaire, administered after the counseling for the test by the counselor. Data were analyzed using mean, percentage, standard deviation and range.

Result

The majority of the women were from urban residences and all of them were literate. Nearly half of the women were in the age group 21-30 years. Mean gestational age was 19.24 ± 2.63 weeks and most were pregnant for the second time. The main reasons for referral were advanced maternal age and abnormal obstetric ultrasonography. The participant's understanding of the test was still incomplete despite the counseling and there was a need for additional counseling/information. One-third of them preferred additional counseling whereas, the remaining two-thirds preferred booklets and pamphlets.

Conclusion

The profiles of pregnant women referred to genetic clinic provide better insight about their background for the health personnel and this study emphasizes rechecking the understanding regarding invasive tests following counseling; which eventually helps for appropriate decision making regarding the tests.

KEY WORDS

Awareness, Decision making, Counseling

INTRODUCTION

Prenatal diagnostic tests have been used for the detection of genetic diseases in a fetus: like Down syndrome which is one of the most common reasons for serious intellectual impairment.[1,2] Prenatal testing range from invasive methods like amniocentesis or chorionic villus sampling (CVS) to recent advances: the non-invasive method like detection of cell-free fetal-deoxyribonucleic acid (DNA) in maternal plasma.[3,4]

The history of amniocentesis and CVS starts from 1877.[5-7] These tests possess a risk for termination of pregnancy by around 0.35% to 1%.[8,9] Indications of prenatal diagnostic tests are advanced maternal age, abnormal screening test results, abnormal obstetric ultrasound scans, prior history of abnormal pregnancy and family history of genetic diseases.[10]

Due to a large population, high birth rate, consanguineous marriage favored in many communities, the prevalence of genetic disorders in India is very high (around 21,400 children with Down syndrome, 9000 with beta-thalassemia and 5200 with sickle cell disease are born every year), which can be detected by prenatal invasive testing.[11,12] The educational and career perspective, economic independence, late marriages, second marriage and awareness on contraceptive methods might have contributed to the delaying of the conception leading to advanced maternal age.[13] These factors might have added to the increasing number of genetic disorders in the newborns in India.

Studies addressing the profile of women who come to seek prenatal invasive testing are scarce. The primary objective of this study is to assess the socio-demographic characteristics and clinical profile of women who are referred for prenatal invasive testing in a tertiary referral center in India.

METHODS

A descriptive, cross-sectional study was undertaken on 60 women who came to the genetic clinic, Department of Pediatrics at All India Institute of Medical Sciences (AIIMS), New Delhi, India for counseling regarding prenatal invasive testing. The duration of this study was 6 months (March 2012 - September 2012). The sample included all pregnant women who were referred to genetic clinics at AIIMS, New Delhi, India for prenatal diagnostic testing and who were willing to participate in the study. The sample size was calculated using formula N= Z2*P*Q/D^2, where N= sample size, Z= Z value i.e., 1.96 for 95% confidence level, P= percentage of uptake of prenatal testing in the previous study, expressed as a decimal (which was 3.9) i.e., 0.039, Q= 1-P, i.e., 0.961, D= the margin of error which is 0.05.[14] Convenience sampling was adopted for the study and a semi-structured questionnaire was administered to the women, which consisted of sociodemographic and obstetric characteristics of women as well as their opinion regarding the prenatal invasive test. During the process of data collection, any doubts or queries arising from the respondent were made clear at the site by the principal investigator. Ethical clearance for the study was obtained from Ethical Committee, AIIMS, New Delhi, India. An information sheet regarding the study was given to the study participants. Informed written consent was taken and confidentiality of the subjects was maintained. Inclusion criteria for sample selection were: pregnant women who had fetal genetic risk factors and were referred to the genetic clinic at AIIMS, New Delhi, India for prenatal diagnostic testing, who could communicate in Hindi or English. For data analysis, descriptive statistical methods included mean, standard deviation, frequency, range and percentage. Data were entered in Microsoft EXCEL and analyzed using SPSS 16.0 version.

RESULTS

The socio-demographic profile of the women attending the genetic clinic (table 1). Nearly half the women were in the age groups 21–30 years followed by 31–40 years age group (46.7% and 43.3% respectively). Interestingly, all the women were literate. Fifteen percent of the women had consanguineous marriage. The occupational status and socioeconomic status were graded as per Kuppuswami's index, where 21.7% of women were skilled workers while 15% of them were professionals. Nearly half of the women belonged to the lower middle class as per Kuppuswami's index.

Table no. 2 depicts the obstetric characteristics of the women. About one-third of the participants reported that the pregnancy was unplanned. A history of a genetic anomaly in the previous child was reported by 16.67% of the participants whereas 20% of them had 2 or more affected children. The mean gestational age of women attending the genetic clinic for prenatal invasive testing was 19.24±2.63 weeks.

Reasons for referral are depicted in figure 1. The common reasons for referral were advanced maternal age i.e., above 35 years (38.3%) followed by abnormal obstetric USG scans (30%).

Table no. 3 illustrates the details of the women regarding prior knowledge/information regarding prenatal invasive testing. The majority of them (78.3%) reported that they didn't have prior knowledge/ information of prenatal invasive tests. Following counseling by the counselor, the majority of the women (71.7%) reported that they were aware of the test. Post-counseling, 70% of them preferred additional information regarding the tests and the majority of them (67.7%) reported that they wanted the information in the form of booklets or pamphlets; while the remaining preferred re-counseling.

Table 1. Socio demographic characteristics of the women (n=60)

Variable	Category	Frequency (%)
Age	< 20 years	2 (3.3)
	21 – 30 years	28 (46.7)
	31 – 40 years	26 (43.3)
	> 40 years	4 (6.7)
Residence	Rural	9 (15)
	Urban	51 (85)
Religion	Hindu	45 (75)
	Muslim	6 (10)
	Christian	6 (10)
	Sikh/others	3 (5)
Education (women)	Primary	4 (6.7)
	Secondary	12 (20.0)
	Higher Secondary	20 (33.3)
	Graduate	14 (23.3)
	Postgraduate and above	10 (16.7)
Education (husband)	Primary	1 (1.7)
	Secondary	1 (1.7)
	Higher secondary	22 (36.7)
	Graduate	22 (36.7)
	Postgraduate and above	14 (23.3)
Type of marriage	Consanguineous	9 (15)
	Non consanguineous	51 (85)
Type of family	Joint	40 (66.7)
	Nuclear	20 (33.3)
Occupation	Unemployed / Housewife	8 (13.3)
	Unskilled	5 (8.3)
	Semiskilled	13 (21.7)
	Skilled	13 (21.7)
	Clerk/shop owner/ farm owner	4 (6.7)
	Semi-professional	8 (13.3)
	Professional	9 (15)
Socioeconomic status	Lower	1 (10)
	Upper lower	6 (40)
	Lower middle	24 (48.3)
	Upper middle	29 (1.7)

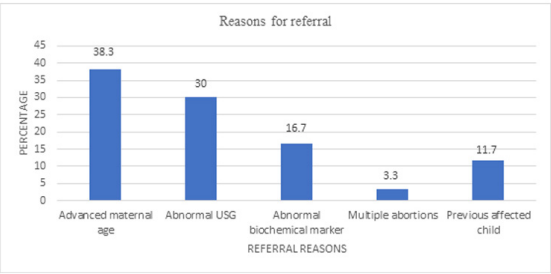

Figure 1. Reasons for referral for the prenatal invasive tests

Table 2. Obstetric characteristics of the women (n=60)

Variable	Category	Frequency (%)
Gravida Mean (2.2 ± 0.68)	1	12 (20)
	2	40 (66.7)
	≥3	8 (13.3)
Prior normal live births	0	9 (15)
	1	3 (5)
	2	40 (66.67)
	≥3	8 (13.33)
Prior still birth(s)	0	48 (80)
	1	10 (16.67)
	≥2	2 (3.33)
Prior pregnancy loss(s)	0	53 (88.3)
	1	4 (6.7)
	≥2	3 (5)
Prior induced abortion(s)	0	56 (93.3)
	≥1	4 (6.7)
Pregnancy planned	Yes	40 (66.7)
	No	20 (33.3)
History of previously affected child (with genetic anomaly)	Yes	10 (16.67)
	No	50 (83.33)
Number of children effected (n=10)	1	8 (80)
	≥ 2	2 (20)
Gestational age (in weeks)	19.24 ± 2.63 (Mean ± SD)	15-23 (Range)

Table 3. Prior knowledge/information regarding the prenatal invasive testing (n=60)

Variable	Category	Frequency (%)
Adequate awareness about the test following counseling	Yes	43 (71.7)
	No	1 (1.7)
	A little bit	16 (26.7)
Presence of prior knowledge/ information of prenatal invasive test	Yes	13 (21.7)
	No	47 (78.3)
Prior source of information about the test (n=13)	Books and internet	7 (55.6)
	Health practitioners (Dr. / Nurse)	6 (44.4)
Need of additional information regarding the testing	Yes	42 (70)
	No	18 (30)
Preferred forms of additional information (n=42)	Booklets and pamphlets	28 (67.7)
	Counseling	14 (33.3)

DISCUSSION

In this study, nearly half (46.7%) of the respondents were of age group 21-30 years. This is contrary to the general assumption that advanced maternal age is the key risk factor for the genetic disorder. This finding is similar to the study done in Netherlands where 48% of women were

below the age of 30 years.[15] The majority of the respondents were Hindu by religion as it is the major religion in India. The majority, (85%) of them were from urban residences which are similar to the study done in Jordan where around 80% of the women were from urban residence.[16] This may be due to easy access to the facility also. Thirty-three percent of the respondents had a higher secondary level of education which is also congruent to the study conducted in the Netherlands but slightly higher than the findings by other studies and was less than that of another study done in the Netherlands.[1,15-17]

Fifteen percent of the marriages were consanguineous which is in contrast to the findings of one of the studies which may be due to consanguineous marriage being common in Muslims which were second to Hindu by religion in our study.[15] The majority of the respondents i.e., 66.7% resided in joint families.

The occupational status and socio-economic status (SES) were graded as per Kuppuswami's index where 21.7% of women were skilled workers while 15% of them were professionals.[18] Nearly half of the women fell into the lower middle class in the SES. In the present study, since a majority of the women were from urban residences, all of them were literate and fell into middle-class family, they had better access to the prenatal invasive tests.

The main reasons for referral in this study were advanced maternal age (38.3%) followed by abnormal obstetric USG scans (30.0%), abnormal biochemical markers, prior pregnancy losses and the previously affected children. These findings are similar to the study done in the USA and Croatia.[19,20] But in Turkey the main reasons for referral were abnormal test results and abnormal ultrasound findings.[10]

In this study, more than two-thirds of the respondents (66%) were pregnant for the second time with a mean gravida of 2.2 which is similar to a study done in the USA.[21] These findings are not congruent with the study done in Turkey.[22] The majority of them (80%) reported no prior stillbirth, which is similar to the study done in Denmark.[23] Only 16.67% respondents reported that they had one stillbirth which is lower than that reported by Mikamo et al.[4] In this study, 6.7% reported that they had one prior pregnancy loss while 5% reported that they had ≥ 2 prior pregnancy losses which are lesser than the findings in Netherlands.[1] The lesser figures of stillbirths and prior pregnancy losses in the present study might be because advanced maternal age followed by abnormal USG were the major reasons for referral in the current study. In other studies, abnormal screening test results, previous history of an affected child and prior pregnancy loss(es)/stillbirths were the main reasons for referral.[1,4,22]

Responding to the history of prior induced termination of pregnancy, 6.7% of respondents reported that they had one induced termination of pregnancy. Nearly one-third (33.3%) reported that the current pregnancy was unplanned which is similar to the study done by Tsai et al.[20] Of the respondents, 16.67% reported a history of a previously affected child with genetic anomaly which is quite higher than the other studies.[24] These obstetric characteristics are congruent with the study findings by Mikamo et al.[4] In this study, the mean gestational age of the respondents was 19.24 ± 2.63 weeks which is quite higher than that of Turkey.[22] This is most likely due to delay in referral or delay in seeking care. The advanced period of gestation (POG) might affect in the decision-making process regarding the termination of pregnancy when required which also adds to mental disturbances and trauma.

The majority of the respondents (78.3%) reported that they didn't have prior knowledge/information regarding prenatal invasive tests. Seventy-one percent of them reported that they were aware of the test following counseling by the counselor. Post counseling, 70% of the respondents preferred to have additional information regarding the tests and the majority of them (67.7%) preferred the information in the forms of booklets and pamphlets. These findings are contrary to the study done in Sweden, where 94% of the participants requested re-counseling.[25] Thus, this study emphasizes re-assessing the post counseling understandings of women undergoing PIT which will ease the decision-making process.

The study tools used were not standardized. A self-developed pretested questionnaire was used.

CONCLUSION

In the present study, the majority of the women were from urban residences, all of them were literate and fell into middle-class families. They also had better access to prenatal invasive tests. Half of the women were below 30 years of age. At the time of genetic counseling mean gestational age was 19.24 ± 2.63 weeks and the majority were pregnant for the second time. The main reasons for referral were advanced maternal age and abnormal USG. The participant's understanding of the test was still incomplete despite the counseling, and there was a need for additional counseling/information. One-third of them preferred additional counseling whereas, the remaining two-thirds preferred booklets and pamphlets.

Implications

This study helps to identify the pregnant women's common risk factors and main reasons for referral to the genetic clinic. It also addresses the need for an assessment of understanding of the invasive tests following counseling and the requirement of further counseling. These profiles provide better insight into the background of pregnant women referred to the genetic clinic for the health personnel to recheck the understanding following counseling.

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