Journal of Advanced Medical and Dental Sciences Research

@Society of Scientific Research and Studies *NLM ID: 101716117*

Journal home page: www.jamdsr.com doi: 10.21276/jamdsr Indian Citation Index (ICI) Index Copernicus value = 100

(e) ISSN Online: 2321-9599; (p) ISSN Print: 2348-6805

# Original Research

## Awareness of prenatal screening for fetal aneuploidy among pregnant women in India

[1]Narita Jamwal, [2]Sumeet Sabharwal, [3]Gagandeep Kour

[1,3]Associate Professor, Department of Obstetrics and Gynaecology, ASCOMS Hospital Sidhra, Jammu, India; [2]Associate Professor, Department of Radiodiagnosis and Imaging, ASCOMS Hospital Sidhra, Jammu, India

***ABSTRACT:***

**Background**: The prenatal screening is an adjunctive tool for detecting any chromosomal abnormalities like aneuploidies in fetus. This is a required asset in medical practice to prevent such conditions that can affect the child as well as their families, mainly the mother. **Aim**: The aim of the study was to assess the awareness of prenatal screening for fetal aneuploidy amongst pregnant Indian women. **Materials and methods**: This questionnaire based study was conducted on 300 pregnant females who reported to the Department of Obstetrics and Gynaecology for antenatal health care. Informed consent was obtained from all the study participants' after obtaining ethical committee approval. Observations recorded were statistically analyzed using Chi-Square statistical tool. Significance was set at 0.05. **Results**: High knowledge regarding prenatal fetal testing was found amongst pregnant females. There was a statistically significant association between trimester (second), education level (graduate), middle social class status and urban settings towards such tests. **Conclusion**: There is high awareness amongst Indian women regarding prenatal screening tests during pregnancy. There is a need for such voluntary program to be undertaken to spread further awareness.

**Keywords**: pre-natal, fetal, screening, pregnant, India

Received: 17 September, 2024 Accepted: 20 October, 2024

**Corresponding author:** Narita Jamwal, Associate Professor, Department of Obstetrics and Gynaecology, ASCOMS Hospital Sidhra, Jammu, India

**This article may be cited as:** Jamwal N, Sabharwal S, Kour G. Awareness of prenatal screening for fetal aneuploidy among pregnant women in India. J Adv Med Dent Scie Res 2024; 12(11):38-42.

## INTRODUCTION

Pre-natal tests for screening of pregnancy are required for identification of genetic disorders in an unborn child. These tests include ultrasound, serum assays and maternal plasma fetal cell-free fetal DNA which detects chromosomal defects such as aneuploidy, micro-deletion and copy number(s) variants. [1] Pre-natal diagnosis has been indicated in women who test positive for genetic disease at screening or there are underlying risk factors like advanced age of mother, family history of any genetic disease/disorder or any abnormal ultrasound observations. [2]

Pre-natal screening tests are utilized for reducing incidence of genetic abnormalities through medical termination of such pregnancies as well as for preventing adverse clinical outcomes for both fetus and mother. [3, 4]

Several nations have already adopted policies for conducting programs for pre-natal fetal testing.[5,6]In India, there are approximately 21,400 children diagnosed with Down's syndrome, 9000 afflicted with beta-thalassemia and 5200 children with sickle cell anemia are born each year. [7, 8]Therefore, nation-wide, government funded prenatal screening program for preventing genetic abnormalities have been recommended as an integral component of routinely performed ante-natal health care. [9] This is essential as an intervention for preventing adverse outcomes on the fetus as well the expectant mother.

Non-invasive pre-natal testing or NIPT is an important step taken in the field of pre-natal care. It was first introduced year, 2011. In less than ten years, this technology is available in over 60 nations and had global market value of 3.9 billion U. S. Dollars in 2019. It is expected to demonstrate growth till 7.3 billion dollars till 2024. [10] Although there are obvious benefits to this screening technique it also has numerous challenges due to different cultural, societal, legal and economic backgrounds.

NIPT can be use mainly for screening of chromosomal abnormalities T21 and T18 in Edwards syndrome, T13 in Patau syndrome and sex chromosomal aneuploidies such as 45,XO in Turner's syndrome, 47,XXY in Klinefelter's syndrome and 47,XYY and 47,XXX in triple X syndrome. This screening tool has high sensitivity in 99.7% (T21), 97.9% (T18), 99.0% in T13, 95.8% in Turner's syndrome and greater than 95% specificity in sex chromosome related aneuploidies. [11] Rarer trisomies, variants in copy number as well as microdeletions like 15q deletion seen in Prader–Willi syndrome and Angelman syndrome, 22q11.2 deletion seen in DiGeorge syndrome, 4p deletion in Wolf–Hirschhorn syndrome, 15p deletion in Cri-du-chat syndrome, and 11q chromosomal deletion seen in Jacobsen syndrome. [12]

However, the sensitivity as well as the positive predictive values (PPVs) for these genetic conditions are significantly less, thus carrying a risk of false positive results while screening for these conditions. [13]

Hence, by keeping in mind such variations this study was designed to assess awareness regarding prenatal screening for fetal aneuploidy among pregnant women in India.

## MATERIALS AND METHODS

a) **Study design and setting:** This was a cross-sectional study conducted at tertiary public hospital. The period of the study was seven months from January 2024 to July 2024.

Ethical approval was duly obtained from Institutional Ethical and Research Committee of the concerned hospital before starting this study.

b) **Study participants and sampling:** Study sample comprised of 300 pregnant females who visited Department of Obstetrics and Gynecology for ante-natal check-up. As there are very few Indian studies conducted on this aspect, it was assumed that 50 percent pregnant females accepted pre-natal screening. With 80% power at 95 % confidence interval and 7% precision, sample size calculated was 296. Hence, the total study sample considered was 300.

c) **Data collection tools and techniques:** For the purpose of collecting data, a self-prevalidated questionnaire was administered to all participants in printed format. The Questionnaire was available in both English and locally prevalent language. Questionnaire items were explained to each of the study participant by investigators assigned with data collection. Questionnaire comprised of 2 components a) Part A collected demographic data like- age of mother, gestational age, parity, consanguinity, genetic disease history in family, educational level and residential address and b) Part B: This part dealt with assessment of acceptance regarding pre-natal screening by using 7 items.

d) **Inclusion criteria:** All pregnant women reporting to the OPD who willingly participated in the study.

e) **Exclusion criteria:** Pregnant women who were unwilling to participate in the study were excluded.

**Statistical analysis:** Data obtained was recorded in percentages. Statistical correlation between socio-demographic features and acceptance was determined using Chi-Square statistical tool.

## RESULTS

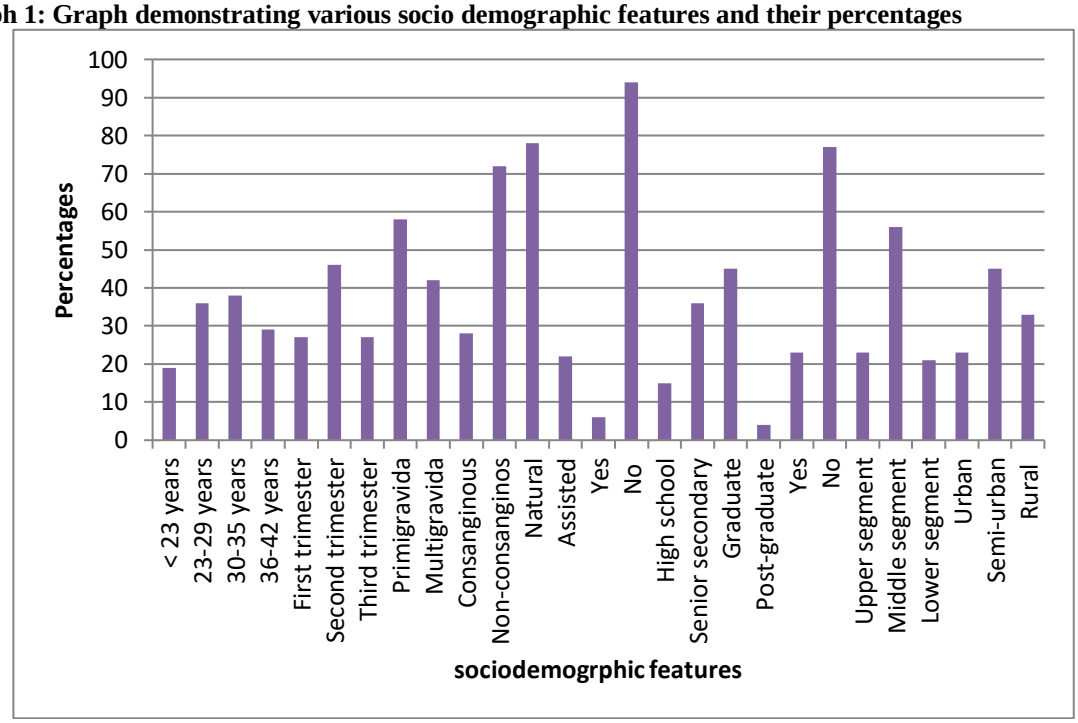
a) **Demographic characteristics:** This questionnaire based study was conducted on 300 pregnant females who visited the Gynaecology and Obstetrics out patient Department of the hospital for routine ante-natal health check-up. The age range of studied population was between 23 to 42 years (median age = 23.5 years). Majority of preganant females who participated in the study were in their second trimester (46%) and 58% were primigravida. 72% of pregnant females were in non-consanguinous marriages. 78% pregnancies were naturally conceived. 94% of participants reported no genetic diseases in the family. Majority of females were graduates (45%) while 77% were not in any employment at time of pregnancy. 56% of studied female participants belonged to middle socioeconomic class while 45% were staying in semi-urban, followed by 33 % in rural and 23% in urban areas (table and graph 1).

**Table 1: Table showing demographic characteristics of studied pregnant females**

| Demographic features | Group distribution | Percentage (%) |
|---|---|---|
| Age at time of study | < 23 years | 19 |
| | 23-29 years | 36 |
| | 30-35 years | 38 |
| | 36-42 years | 29 |
| Gestation period | First trimester | 27 |
| | Second trimester | 46 |
| | Third trimester | 27 |
| Parity | Primigravida | 58 |
| | Multigravida | 42 |
| Consanguineous status | Consanginous | 28 |

| | | |
|---|---|---|
| | Non-consanginos | 72 |
| Conception type | Natural<br>Assisted | 78<br>22 |
| Is there a family history of any Genetic disorder | Yes<br>No | 06<br>94 |
| Level of education | High school<br>Senior secondary<br>Graduate<br>Post-graduate | 15<br>36<br>45<br>04 |
| Employed | Yes<br>No | 23<br>77 |
| Socioeconomic status | Upper segment<br>Middle segment<br>Lower segment | 23<br>56<br>21 |
| Type of residence | Urban<br>Semi-urban<br>Rural | 23<br>45<br>33 |

**Graph 1: Graph demonstrating various socio demographic features and their percentages**




**b) Knowledge:** 89% of studied pregnant female population responded in affirmation regarding undergoing prenatal screening test if advised by a Gynaecolgist, 03 % were not sure about getting tested and 09 % did not want to undergo any genetic tests. 89 % were willing to undergo specialized tests if any abnormal findings were elicited in ultrasound and blood investigations. 98% of studied participants were willing to discuss any abnormalities detected with genetic counselors and healthcare professionals.

**c) Acceptance and precipitation:** 78% of studied population was having the acceptance of the idea about the requirement of a government led program for ante- and pre-natal screening during pregnancy.

On applying Chi-square tests for determining association between sociodemographic features and acceptance, a highly significant P value (0.05) was obtained. Pregnant feamloes in age range of 35 to 45 years demonstrated highest probability of acceptance of these genetic screening tests (P=0.04). Employment and socioeconomic status were found to have statistically significant association (P= 0.04 and 0.05, respectively.

## DISCUSSION

The prenatal screening tests play an important role in identifying the genetic diseases. Once identified these tests pave the path towards interventions for preventing as well as managing any adverse results for both fetus as well as mother. Since making a decision is emotionally difficult, most expectant

couples seek the advice of a Gynaecologist and/or genetic counselor in these conditions.

Ogamba et al (2021) in their study in Lagos reported that a statistically significant study population i.e., 81.3% had a positive attitude regarding conducting screening tests for expectant females.

75.1% of studied population indicated their willingness in undergoing diagnostic tests in case there were concerning findings in ultrasound or any other investigations used for screening pregnancy. [14]

Similar studies conducted in Greece, Ibadan and Sokoto have reported significantly high acceptability levels for prenatal screening tests with 68%, 75 % and 95.2%, respectively. [15, 16, 17] Arafah et al (2021) conducted their study on Saudi Arabian participants who expected the government to operate facilities for providing genetic screening and counseling to their citizens. [18]

All the studies cited above are in support to the present study findings conducted on Indian pregnant women who expressed significantly high awareness regarding prenatal screening when correlated with socio demographics parameters studied. Women belonging to higher age groups and education level were more supportive of these screening tests. In a case scenario, if there is an active prenatal genetic screening testing facility which is government led, most of the study participants were willing to utilize it if indicated or advised by their doctors. As the stage of pregnancy progresses, significantly higher numbers of women become receptive towards getting their fetus examined for any abnormalities. Our findings have been supported by findings of a study conducted by Arumugam et al (2024) in Andhra Pradesh. [19]

Noninvasive pre-natal screening is a genomic technique which detects cell-free placental DNA which is in circulation in mother's blood. In this technique, parallel DNA sequencing of maternal as well as placental/fetal DNA fragments is done simultaneously. Sequencing performed is random in nature, targeted towards single nucleotide polymorphisms (SNPs). [20] Sequencing can also be done by measurement of hydrogen ions release when nucleotides get added to DNA template. This process is called as semi-conductor sequencing. [21] Use of microarray technique is also done for quantification of DNA. Both the health-care professionals as well as patients experience marketing practices, rapid evolution in practice guidelines and incorporation of prenatal screening in obstetrics practice. Hence, there is a constant need for such government led programs especially in India where most of the population cannot afford expensive testing tools.

## CONCLUSION

The prenatal genetic screening is of importance as it saves severe outcome for both the fetus as well as their families. A high acceptance and awareness in the studied population in India demonstrates the significant requirement for wide availability of this facility pan India. These facilities preferable should be government run under specific practice guidelines for appropriate outreach to those indicated.

## REFERENCES


1. Carlson LM, Vora NL. Prenatal diagnosis: screening and diagnostic tools . Obstet Gynecol Clin North Am 2017;44:245-56.
2. Caceres V, Murray T, Myers C, Parbhoo K. Prenatal genetic testing and screening: a focused review . Semin Pediatr Neurol 2022, 42:100976-90.
3. Evans MI, Andriole S, Evans SM. Genetics: update on prenatal screening and diagnosis . Obstet Gynecol Clin North Am. 2015, 42:193-208.
4. Gaboon NE, Bakur KH, Edrees AY, Al-Aama JY. Attitude toward prenatal testing and termination of pregnancy among health professionals and medical students in Saudi Arabia. J Pediatr Genet 2017;6:149-54.
5. Gadsbøll K, Petersen OB, Gatinois V. Current use of noninvasive prenatal testing in Europe, Australia and the USA: a graphical presentation. Acta Obstet Gynecol Scand 2020;99:722-30.
6. Phadke SR, Puri RD, Ranganath P. Prenatal screening for genetic disorders: suggested guidelines for the Indian Scenario. Indian J Med Res 2017;146:689-99.
7. Verma IC, Bijarnia S. The burden of genetic disorders in India and a framework for community control. Community Genet 2002;5:192-6.
8. Verma IC, Saxena R, Kohli S. Past, present & future scenario of thalassaemic care & control in India . Indian J Med Res 2011;134:507-21.
9. Massa JD, Arora V, Lallar M. Current status of noninvasive prenatal testing and counseling considerations: an Indian perspective. J Fetal Med 2019;7:9-16.
10. Marcon AR, Ravitsky V, Caulfield T. Discussing non-invasive prenatal testing on Reddit: the benefits, the concerns, and the comradery. Prenat Diagn 2021; 41:100–10.
11. Gil MM, Accurti V, Santacruz B, Plana MN, Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. Ultrasound Obstet Gynecol 2017;50:302–14.
12. Hui L, Bianchi DW. Noninvasive prenatalDNA testing: the vanguard of genomic medicine. Annu Rev Med 2017; 68:459–72.
13. Meredith S, Kaposy C, Miller VJ, Allyse M, Chandrasekharan S, Michie M. Impact of the increased adoption of prenatal cfDNA screening on non-profit patient advocacy organizations in the United States. Prenat Diagn 2016; 36:714–19.
14. Ogamba CF, Babah OA, Roberts AA, Nwanaji-Enwerem JC, Nwanaji-Enwerem P, Ikwuegbuenyi CA, Ologunja OJ. Knowledge, attitudes, and decision making towards prenatal testing among antenatal clinic attendees in Lagos University Teaching Hospital: an institution-based cross-sectional study. Pan Afr Med J 2021;39:106-10.
15. Karagkiouzis T, Sifakis S, Makrithanasis P. Awareness of prenatal screening for fetal aneuploidy among pregnant women in Greece. In Vivo 2015;29:155-60.
16. Adekanbi AO, Olayemi OO, Fawole AO: The knowledge base and acceptability of prenatal diagnosis

by pregnant women in Ibadan. Afr J Reprod Health 2014;18:127-32.
17. Ahmed Y, Panti AA, Umar AG et al. Knowledge and acceptability of prenatal diagnosis among pregnant women attending antenatal clinic in a tertiary health institution in Sokoto, Nigeria. Int J Reprod Contracept Obstet Gynecol 2021;10:3678-83.
18. Arafah A, Al Jawadi MH, Aldheefi M, Rehman MU. Attitude and awareness of public towards genetic testing in Riyadh, Saudi Arabia. Saudi J Biol Sci 2021;28:255-61.
19. Arumugam S, Kalluri S, Sharmila V. Understanding the Awareness of Prenatal Genetic Screening Tests Among Pregnant Women in India: A Cross-Sectional Study. Cureus 2024; 16(3): e56932-40.
20. Palomaki GE, Kloza EM, Lambert-Messerlian GM. DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genet Med 2011;13:913–20.
21. Liao C, Yin AH, Peng CF. Noninvasive prenatal diagnosis of common aneuploidies by semiconductor sequencing. Proc Natl Acad Sci USA 2014;111:7415–20