



## GENETIC ABNORMALITIES IN PRENATAL TESTING AMONG MATERNAL WOMEN

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## Abstract

## Background:

Pregnancy is a astounding journey bursting with happiness and expectation. Though, it can similarly be a period of indecision and apprehension, particularly when it emanates to genetic difficulties. Genetic complications can arise during pregnancy, foremost to probable health problems together the baby and the mother. Genetic problems can ascend due to a variability of issues, with inherited gene mutations, abnormalities in chromosomes, and environmental effects. Inherited gene mutations, which are crossed down from paternities to their offspring and can upsurge the risk of positive genetic syndromes. This study supports respected perceptions to the unending treatise, anticipating the penchant for aggressive prenatal screening technique to ensure comprehensive and ethically sound approach to prenatal care in the unique socio-cultural landscape of India.

## Methods:

A cross-sectional study design carried out over a span of 12 months at a selected tertiary care hospitals in India. A total of 190 pregnant women who underwent invasive prenatal testing via amniocentesis alongside with 95 partners participated in the research were selected through convenience sampling. To ensure a representative sample, participants were recruited from diverse demographic backgrounds

## Results:

The greater part of members, 80% of expecting ladies and 75% of companions were mindful of hereditary testing choices. Amniotic fluid examination emerged as the most perceived strategy, with 70% of expecting ladies and 65% of companions learned about the system. *Conclusion:* 

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Keywords: Amniocentesis; Genetic disorders, Inheritance, Pregnancy, Social Impact.

## 1.Introduction

Prenatal evaluation and care in general is attributed as a truly remarkable changein recent times, with a promise towards a healthier pregnancies alongside with profoundnewer understandings on the genetic profiles of the neonates during their developmental stage itself (Rappaport.,2008; Carlson &Vora., 2017). Genetic conditions such as thalassemia and Down syndrome contributes significantly when on considering India's prevalence in genetic disorders, with rising statistics each year (Gajbhiye &Gaitonde.,2022). With advanced prenatal genetic testing has been an intersecting element for India's deeply diverse cultural fabric, a unique set of considerations that are emerging in present day scenario (Chattoo., 2018). Indian expectant parents are facing the psychological burden regarding fetal abnormalities via prenatal testing and were in the dire situation of grappling with the complex social dilemmasand ethical beliefs (Allyse et al, 2015). Cultural diversity followed by long-held values having profound impact concerning on the ethical discussions revolving around the current practices of prenatal testing in India (Haidar et al., 2018). In such an intricate climate, the influence of invasive techniques like amniocentesis become quite crucial (Likaret al., 2020).

Amniocentesis is an invasive method employed primarily for extraction of amniotic fluid from fetus using specialized needle and the extract was further then subjected for screening of any potential abnormalities. Despite the technique initially was introduced in 1950s as a means for genetic screening, their clinical significance gained immense interest in recent times (Kazal et al., 2021). From the pioneering work of Serr and colleagues for extraction of amniotic fluid samples primarily for determination of fetal sex (Theodosiou, & Johnson, 2011). Further progress were made by Steele and Breg in 1977 utilizing amniocentesis as an essential means for cultivation of amniotic cells and analysis of the karyotypes, laying the very foundation towards prenatal assessment. With growing time, amniocentesis has gained immensely as a widely relied approach in identification of numerous disorders, most notably on some of the prominent chromosomal anomalies that are quite observable from during the initial stage of the fetal development. With the modernized practices that are involved with collection of amniotic fluid in second trimester to perform a well-throughput assessment on the fetal genetics offering better safety measures with accuracy compared with the real-time ultrasound monitoring which is most commonly employed screening approach on potential issues (Ciortea et al., 2023). The advancements concerning pre-birth screening posed ongoing challenges for obstetricians supporting pregnant women. By gaining access under such test is crucial and is much more important for expectant mothers who is seeking for prenatal assessment of genetic conditions that impact seriously with the health and development of fetus (Bansal, & Jhaveri, 2022).

This research aimed at investigating on how the genetic anomalies are identified using the prenatal screening and how it resonates from the social standpoint and especially from an India's diverse societal weave, emphasized roles on probing tests. Indian subcontinent, a mixture of faiths, traditions and wellness globally, offering unique phase to explore the key impacts of delving the prenatal exams. Expectant mothers as well as fathers who are confronting this nuanced milieu—particularly whether towards experiencing with the probing screening pattern—standing as nexus for the examination. We seek their stories to comprehend navigating desires for children's health and respecting cultural customs, often pursuing both with probing tests bringing peace or distress. This study provides deeper insights and social responses from the study participants for exploring the key trends and sensitive policies concerning genetic abnormalities in prenatal testing.

## 2. Materials and Methods

## 2.1.Study Design

This study looks back at prenatal screening results to find out how often different genetic issues occur in Indian pregnancies at a big hospital. The medical records provide a lot of details about the rates of various conditions identified before birth.

## 2.2.Data Source

Medical Records: Having records from a specific time that meet the criteria of including data from prenatal exams would enable detailed analysis. A large number of cases would provide strong statistical support to understand the types of abnormalities found through prenatal screening in selected tertiary care hospitals in India.

## 2.3.Study Participants

Inclusion Criteria: Allowing access to patient files and digital health records from prenatal centers nationwide would help in the review process.

## 2.4.Sample Size

Gather information from a wide range of prenatal cases across various regions and demographics in the area, specifically those admitted to tertiary care hospitals. This diverse group of patients will enable researchers to examine different experiences and social factors in detail.

## 2.5.Variables Examined

To observe and understand on the social impacts due to genetic abnormalities that are examined via prenatal testing was recorded. The collected data comprised of several key variables, each of which plays a pivotal role in shaping the landscape of prenatal care and its ethical and societal implications.

## 2.6. Amniocentesis procedure

The procedure is typically conducted after the 15th week of pregnancy, ideally after the 16th week, to minimize potential complications. Once the prerequisites are confirmed and preparations are complete, the procedure begins with the introduction of a 20-gauge or 22-gauge spinal needle into the amniotic cavity, guided continuously by ultrasound. Emphasis is placed on achieving a firm entry to prevent the tenting of the amniotic membrane.

Upon confirmed entry into the amniotic cavity, amniotic fluid is slowly aspirated. The initial 1 ml to 2 ml is discarded due to its higher likelihood of maternal cell contamination. Approximately 18 ml to 20 ml of amniotic fluid is then extracted for karyotype testing. The needle is carefully withdrawn once an adequate amount of amniotic fluid has been obtained.

It is generally advised to avoid entry into the amniotic cavity through the placenta to minimize the risk of a bloody tap, particularly in Rh-negative women. Besides amniotic fluid collection, 5 ml maternal blood was simultaneously drawn as a means for ruling out any maternal cell contaminations. This methodology ensured precise and fastidious implementation of processes, entailing considerations such as needle option, timing, and fluid amount employed for testing. Continuous ultrasound guidance played pivotal role towards augmenting safety and accuracy of amniocentesis procedure.

## 2.7.Type of Genetic Abnormality Detected

Detection of any genetic abnormalities is the key element and serves as the foundation for the research. This includes recording the outcomes of prenatal screenings showing genetic issues. The issues in general are Down Syndrome to neural tube defects and other chromosomal disorders like Patau syndrome, Edwards syndrome, and Triploidy XXY. Upon knowing how common the genetic anomalies and its impacts provide necessary details on difficulties and challenges faced by expectant parents and its wider impacts on the society.

## 2.8.Maternal Age at the Time of Testing

Maternal age serves as a crucial factor when subjected for prenatal screening. A woman's age at the time of the genetic evaluation tend to greatly influence with their options/ preferences. This is particularly due to its impact on decisions on whether to have prenatal exams and what to do with its outcomes. Upon closer examination at the maternal age statistics aided us to understand the thoughts and trends governing with the genetic tests.

## 2.9.Gestational Age at the Time of Testing

The timing for conducting genetic screening for pregnancy serves as very crucial factor since it affects choices available and result accuracy as well. We tracked expecting parents had genetic tests done during pregnancy. The information aided in understanding how the timing influencedecision-making and how it leads for worse or better outcomes from the prenatal tests conducted.

## 2.10. Pregnancy Continuation or Termination Based on Test Results

One major factor when on considering the current investigation is with whether the pregnancy is continued or ended on the basis of the achieved outcome from the prenatal investigations. In order for grasping and understanding the real-life impacts concerning with the prenatal screening practice is that we need to the choices made by expectant parents after receiving the prenatal findings. By analysis of such decisions revealed ethical and social impacts with regards to the influence of genetic testing.

## 2.11. Survey Development and Distribution

To explore social impacts on the genetic abnormalities identified via prenatal tests in India, detailed questionnaire survey wascreated after reviewing on the relevant investigations and identification of key themes as well as questions from previous literature sources. Before sharing the survey questions, information pamphlet was given as a means to inform the study participants concerning the various testing options that are available for conducting prenatal testing during pregnancy. The options comprised with invasive methods such as amniocentesis and safer alternative like non-invasive prenatal test and maternal serum screening (Bansal & Jhaveri, 2022; Rose et al., 2022). This pamphlet provided detailed overview on the procedures, best period to conduct testduring pregnancy, accuracy rates, likely risks, and whether tests that

were offered for screening or definitive diagnoses. Participants received brief descriptions of possible test outcomes and implications for pregnancy. The study aim was towards better understanding on how the genetic abnormalities revealed prenatal investigations and how it influences cultural and social decisions of expectant Indian mothers and partners.

## 2.12. Survey Timeline and Language Options

Detailed survey conducted during long period, starting since the latter half of 2019 and continuing till 2020. Questionnaires created in both English and main local dialect, as a means to include the majority of the respondents (citizens) who are speaking at different languages. The approach aimed to reach people across various language backgrounds and provided complete picture of residents' opinions who are from diverse linguistic roots.

## 2.13.Recruitment and Data Collection

Recruitment for survey was conducted at well-known Medical Centre of Delhi, serving as main research site for data collection. Expecting fathers and mothers were met and were selected on the basis of enrolment criteria (being currently pregnant and willingness for participating in the research), were invited during their regular prenatal checkups. Joining were optional, and all of its participants provided informed consent before sharing its opinions. In addition, certain participants chosen for discussing on how the cultural influences as well as societal expectations affecting pre and post-natal care decisions via conducting detailed interviews.

## 2.14. Ethics Approval

Ethical approval strictly was solely obtained from the testing of amniotic fluid via private organization 'Lifecell' for a period of one whole year. Through imparting strict ethical review ensures that the research conducted appears to adhere well with every ethical guideline whilst also safeguarding all the participant's privacy and rights.

## 2.15.Data Analysis

In order to explore on the potential effects from the study population and their perspectives towards invasive amniocentesis procedure, the current research has analysed survey responses on the basis of several factors like: socioeconomic characteristics, stated intentions such as usage of outcomes, and participants' knowledge on the genetic conditions plus accessible prenatal screening options within Indian settings. Intricate statistical examination was executed employing an assortment of tests such as Pearson Chi-Square analyses, Kruskal-Wallis along with Mann-Whitney U tests, plus Kendall's tau correlations where applicable. Analysis was conducted employing particular statistical software program (e.g., IBM SPSS Version.24).

## 2.16.Threshold of Statistical Significance

The data we gathered required stringent standards to account for multiple comparisons, using a threshold of p < 0.005 to minimize false positives and yield reliable conclusions.

## 2.17. Presentation of Quantitative Results

Quantitative findings are presented in the form of frequencies of responses, expressed as percentages (%). This approach allows for a clear and concise presentation of the survey results, enabling readers to gain insights into the perspectives and concerns of the expectant parents surveyed.

## **3.RESULT**

## **3.1.Participants' Demographics**

Demographic variables of patients provide a comprehensive view of the study population and offer crucial insights into the factors that might influence awareness, perception, and decision-making regarding prenatal genetic testing. Understanding these demographics helps in assessing how different social and economic backgrounds affect the accessibility, acceptance, and willingness to undergo genetic screening.

## **3.2.Age Distribution and Its Implications**

The study sample comprises individuals across various age groups, ensuring a balanced representation of reproductive age demographics. The highest percentage of respondents falls within the 25–30 years category (24.2%), followed by the 18–24 years group (20.7%). Participants aged 31–35 years and 36–40 years both account for 18.6% of the sample, while those above 40 years constitute 17.9% (Table 1, Figure 1). This age distribution reflects a normal reproductive age range, emphasizing that the majority of participants are at a stage where prenatal genetic testing is most relevant. The relatively even distribution across age brackets enhances the study's ability to analyse awareness and decision-making variations among different age groups.

Age (in years)		
	Frequency	Percent
18 - 24 years	59	20.7
25 - 30 years	69	24.2
31 - 35 years	53	18.6
36 - 40 years	53	18.6
Above 40 years	51	17.9
Patient group		
	Frequency	Percent
Pregnant women	190	66.7
Partners	95	33.3
Educational Level		· · · · · · · · · · · · · · · · · · ·
	Frequency	Percent
No formal education	44	15.4
Primary school	57	20.0
High school	64	22.5
Undergraduate degree	59	20.7
Postgraduate degree	61	21.4
Employment Status		· · · · · · · · · · · · · · · · · · ·
	Frequency	Percent
Employed (Full-time)	77	27.0
Employed (Part-time)	57	20.0
Homemaker	76	26.7

## Table 1: Participants' demographics

International Journal of Innovation Studies 9 (1) (2025) 75 Unemployed 26.3 Socioeconomic Status (Monthly Household Income in INR) Frequency Percent Less than Rs. 10,000 74 26.0 79 Rs.10,000 - Rs.25,000 27.7 Rs.25,001 - Rs.50,000 87 30.5 More than Rs.50,000 45 15.8 Residence Frequency Percent 157 Rural 55.1 Urban 44.9 128 Total 285 100.0

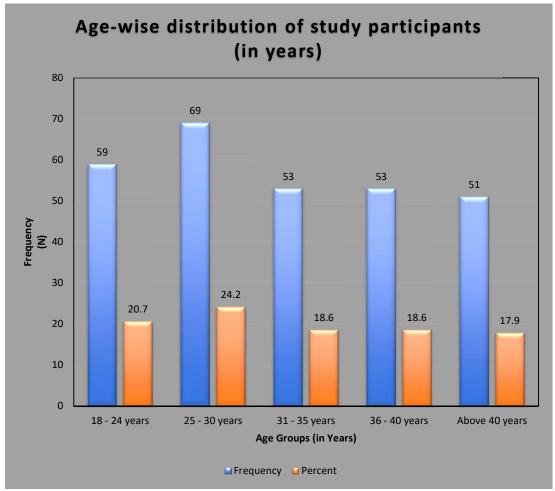


Figure 1. Age-wise distribution of study participants (expectant mother & partners) 3,3,Patient Group:

To ensure a holistic understanding of the social impact of prenatal genetic testing, the study includes both pregnant women and their partners. Pregnant women constitute the majority

(66.7%), while partners represent 33.3% of the sample (Table 1, Figure 2). The inclusion of partners is critical, as they play an integral role in decision-making, providing emotional support, and addressing the outcomes of prenatal screening. This balanced inclusion ensures that perspectives from both maternal and paternal figures are captured, leading to a more comprehensive assessment of social and familial concerns associated with genetic abnormalities.

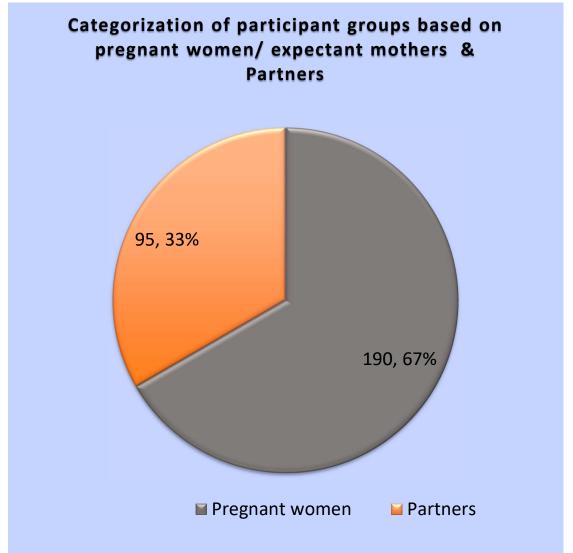


Figure 2.Distribution of study participants based on expectant mothers & partners

## **3.4.Educational Influence on Awareness**

Education significantly influences awareness and decision-making regarding prenatal genetic testing. The study includes participants from various educational backgrounds: the largest group (22.5%) possesses a high school education, followed by postgraduate degree holders (21.4%) and undergraduate degree holders (20.7%). Approximately 20% have completed primary school, while 15.4% have no formal education (Table 1, Figure 3). This diverse educational distribution enables an analysis of how knowledge and awareness levels correlate with educational attainment. Higher education levels are likely to be associated with greater

knowledge about prenatal genetic testing, whereas lower education levels might necessitate enhanced awareness programs and accessible counseling services to improve understanding and informed decision-making.

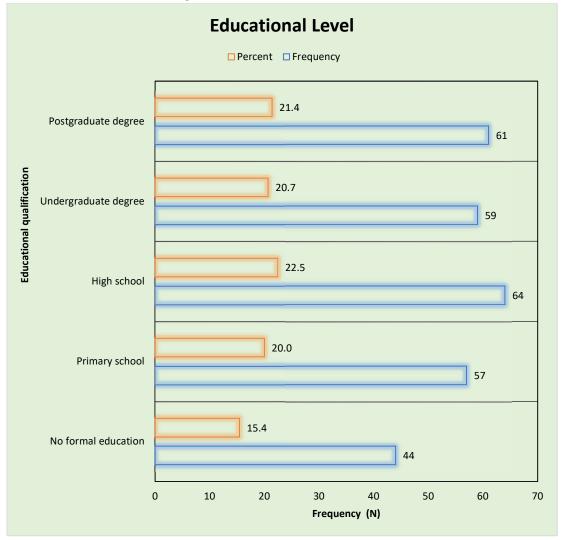
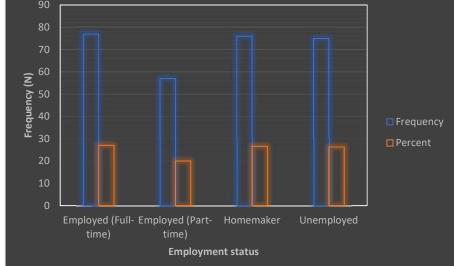


Figure 3. Educational level distribution among study participants (partners and expectant mothers),

highlighting the varying levels of formal education

## 3.5.Employment Status and Financial Dependency in Decision-Making

The employment status of participants is evenly distributed, with full-time employees constituting 27% and part-time employees 20%. A significant portion of respondents are homemakers (26.7%) and unemployed individuals (26.3%) (Table 1, Figure 4). A high number of homemakers and unemployed participants indicates financial dependence, which may impact their ability to access healthcare services, including prenatal genetic testing. Assessing employment patterns helps identify socioeconomic constraints that may affect healthcare



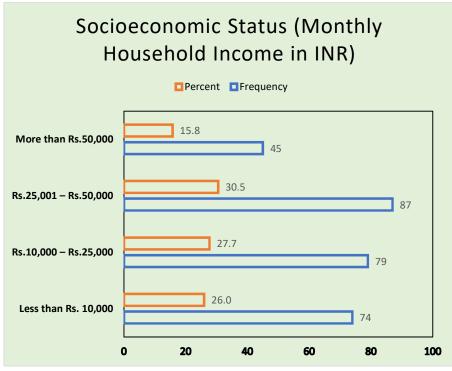
choices and influence the accessibility of genetic testing services for expectant mothers and

their partners.

**Figure 4.** Employment status distribution among study participants (partners and expectant mothers), illustrating variations in full-time employment, part-time employment, homemaking, and unemployment.

## 3.6.Socioeconomic Status and Its Impact on Healthcare Accessibility

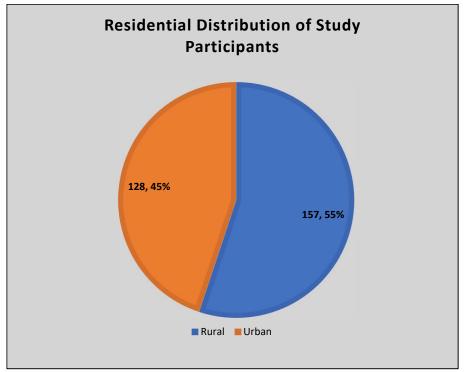
Socioeconomic status, represented by monthly household income, is a crucial determinant of healthcare accessibility and decision-making. The majority of participants (30.5%) belong to the income group of Rs. 25,001–Rs. 50,000, followed by 27.7% in the Rs. 10,000–Rs. 25,000 range. Around 26% fall under the lowest income category (< Rs. 10,000), while 15.8% earn more than Rs. 50,000 per month (Table 1, Figure 5). The significant proportion of low-income participants suggests that financial constraints may be a barrier to accessing prenatal genetic testing. Higher-income groups might have greater access to advanced healthcare services, influencing their perspectives and decisions regarding genetic screening. Addressing financial disparities through subsidized healthcare programs and educational initiatives can improve the accessibility of prenatal testing.



**Figure 5.** Socioeconomic status distribution among study participants (partners and expectant mothers), categorized by monthly household income (INR).

## 3.7. Residence and the Rural-Urban Divide in Healthcare Accessibility

The majority of participants reside in rural areas (55.1%), while 44.9% belong to urban regions (Table 1, Figure 6). This rural predominance highlights the need to evaluate awareness and accessibility of prenatal genetic testing in less developed areas, where healthcare facilities might be limited. Disparities in medical infrastructure, cultural beliefs, and availability of genetic counselling services between rural and urban settings can significantly impact the acceptance and utilization of genetic screening. Understanding this divide can aid in developing targeted interventions to enhance healthcare outreach and improve prenatal screening accessibility in underserved regions.



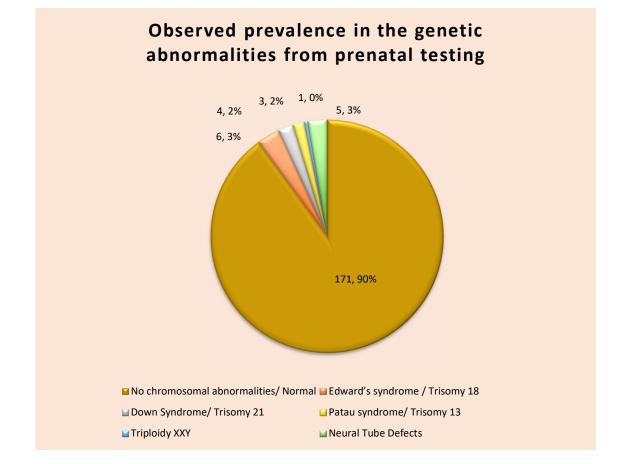
**Figure 6.** Residential distribution of study participants (partners and expectant mothers) across urban and rural areas, highlighting potential disparities in access to prenatal genetic testing.

## **3.8.Genetic Findings from Amniocentesis:**

Study participants showed no signs of fever, unusual vaginal bleeding or chorioamnionitis. No instances of fetal injury were reported during the procedure in any of the cases. For all patients identified with a chromosomally abnormal fetus, a second trimester abortion was conducted at the healthcare centre following comprehensive counselling sessions with the patients. Results obtained from AF sample revealed comprehensive spectrum of genetic findings in the studied population (Table 2). The majority, constituting 90%, exhibit no chromosomal abnormalities, indicating a reassuringly high proportion of normal fetal development. However, the presence of genetic anomalies is notable, with Edward's syndrome (Trisomy 18) identified in 3.16% of cases, Down Syndrome (Trisomy 21) in 2.10%, and Patau syndrome (Trisomy 13) in 1.58% (Figure 7.). Signs of Trisomy complications indicates chromosomal abnormalities associated with distinct clinical features and potential health challenges. Additionally, Triploidy XXY is identified in 0.53% of cases, suggesting an extra set of chromosomes. Neural Tube Defects, accounting for 2.63%, further contribute to the complexity of genetic variations detected. This comprehensive breakdown underscores the significance of AF test as a diagnostic tool, providing valuable insights into the genetic landscape of the studied population and offering critical information for informed medical decision-making during pregnancy.

in the study population		
Result	Frequency (N)	%
No chromosomal abnormalities/ Normal	171	90
Edward's syndrome / Trisomy 18	6	3.16
Down Syndrome/ Trisomy 21	4	2.10
Patau syndrome/ Trisomy 13	3	1.58
Triploidy XXY	1	0.53
Neural Tube Defects	5	2.63

 
 Table 2: Observed prevalence in the genetic abnormalities from prenatal testing in the study population



# Figure 7. Frequency distribution of participants on genetic abnormalities from prenatal testing

## among the study population

Understanding the awareness and knowledge of genetic testing is crucial in evaluating how well-informed maternal women and their partners are about prenatal screening options and their implications. The responses from participants (**Table 3**), provided key insights into their familiarity with genetic testing procedures, highlighting variations in awareness levels between the two groups.

The majority of both maternal women and their partners reported being aware of genetic testing, with 80% of maternal women and 75% of partners indicating prior knowledge. The

slight disparity suggests that maternal women, who undergo routine antenatal visits, may have greater exposure to healthcare professionals and educational resources related to genetic testing. Nonetheless, the high level of awareness among partners underscores the increasing role of paternal involvement in prenatal decision-making.

Responses	Maternal Women (N = 190)	Partners (N = 95)	
Awareness on Genetic Testing	152 (80%)	71 (75%)	
Familiarity with Prenatal Test	ing Types		
- Amniocentesis	143 (75%)	66 (70%)	
- Maternal Serum Screening	114 (60%)	52 (55%)	
- NIPT	133 (70%)	62 (65%)	
Perceived Accuracy of			
Tests	Maternal Women (N = 190)	Partners (N = 95)	
- High	85 (45%)	47 (50%)	
- Moderate	57 (30%)	28 (30%)	
- Low	38 (20%)	19 (20%)	

 Table 3: Participants' responses on 'Observed Awareness and Knowledge of Genetic

 Testing'

## **3.9.**Familiarity with Prenatal Testing Types

Among the different types of prenatal genetic tests, amniocentesis was the most widely recognized, with 75% of maternal women and 70% of partners indicating familiarity with the procedure. This aligns with the historical use of amniocentesis as a standard diagnostic test for chromosomal abnormalities. Meanwhile, maternal serum screening had the lowest recognition, with only 60% of maternal women and 55% of partners aware of the test. This could indicate a need for greater public education on the role of biochemical markers in prenatal screening. Interestingly, non-invasive prenatal testing (NIPT) was known by 70% of maternal women and 65% of partners, reflecting growing awareness of newer, less invasive screening techniques. The relatively high awareness of NIPT suggests increasing adoption and accessibility of this technology in prenatal care.

## **3.10.Perceived Accuracy of Genetic Tests**

Perceptions of test accuracy varied among participants, with 45% of maternal women and 50% of partners considering genetic tests to be highly accurate. A moderate level of confidence was reported by 30% of maternal women and partners, while 20% of maternal women and partners perceived the accuracy as low. These perceptions may be influenced by misconceptions, past experiences, or healthcare guidance. The slightly higher confidence in test accuracy among partners suggests that men may have a more optimistic perception of medical diagnostics, possibly influenced by scientific literacy or discussions with healthcare providers.

## **3.11.Implications for Prenatal Counseling**

The findings suggest a generally high awareness of genetic testing among both maternal women and their partners, yet gaps remain in the depth of knowledge regarding specific screening methods. The relatively lower awareness of maternal serum screening compared to

amniocentesis and NIPT indicates a need for targeted educational efforts. Moreover, while many participants viewed genetic tests as highly accurate, a significant portion remained sceptical, highlighting the importance of comprehensive prenatal counseling to address concerns, clarify test reliability, and ensure informed decision-making.

By identifying these awareness and knowledge trends, healthcare providers can develop tailored strategies to improve prenatal genetic education, ultimately enhancing the decision-making process for expecting parents.

#### 3.12. Chi-square test: Education Level and Knowledge about Genetic Testing

A chi-square test was conducted to determine whether there was an association between educational attainment and prior awareness of genetic testing before pregnancy(**Table 4**). The results revealed a significant association ( $\chi^2 = 10.544$ , p = 0.032), indicating that knowledge of prenatal genetic testing correlates with one's educational background. Among participants with no formal education, 59.1% (26 out of 44) were aware of genetic testing, whereas 40.9% (18 out of 44) had no prior knowledge. A contrasting trend was observed among those with primary education, where 59.6% had no awareness and only 40.4% were informed about genetic testing before pregnancy.High school graduates showed a notable improvement in awareness levels, with 67.2% (43 out of 64) indicating prior knowledge of genetic testing. The trend continued among participants with undergraduate (49.2%) and postgraduate (47.5%) degrees, suggesting that as individuals receive more scientific education and health-related exposure, their awareness of genetic screening increases.

		about	Knowledge about Genetic Testing Before			
		Pregna		Total		
		No	Yes		Pearson Chi-square	p-value
Educational Level	No formal education	18	26	44		
	Primary school		23	57		
	High school	21	43	64	10.544	0.032
	Undergraduate degree	30	29	59	10.344	0.032
	Postgraduate degree		29	61		
Total		135	150	285		

Table 4 Chi-square test:	Education <b>k</b>	evel and K	Knowledge	about genetic	c testing
		• • • • • • • • • • •			

## 3.13.Chi-square test: Primary Decision-Making and Stress Factors

A chi-square test was performed to explore the relationship between the primary decisionmaker regarding amniocentesis and the level of stress experienced during the decision-making process(**Table 5**). The results showed a statistically significant association ( $\chi^2 = 18.801$ , p = 0.027), highlighting that decision-making authority plays a crucial role in influencing stress

levels in pregnant women and their partners. Among those who made the decision independently, 35.7% (25 out of 70) reported no stress, while 24.3% (17 out of 70) found the decision highly stressful. This suggests that self-decision-making may provide a sense of control and reduce anxiety. However, those whose partners made the decision experienced higher stress levels, with only 17.9% (14 out of 78) reporting no stress, whereas 34.6% (27 out of 78) found it moderately stressful, and 20.5% (16 out of 78) considered it very stressful. When doctors were the primary decision-makers, fewer participants (19.0%) reported no stress, while 30.2% found the decision moderately stressful, and another 30.2% considered it very stressful, indicating that clinical recommendations may add pressure and reduce personal autonomy. **Table 5: Chi-square test: Primary decision making and Stress factors** 

			ressful v amniocer	vas the dec ntesis?	cision to			
		Not stressf ul at all	Slightl y stressf ul	Moderatel y stressful	Very stressf ul	Tota 1	Pearson Chi- square	p- value
Who	Self	25	12	16	17	70		
was the	Partner	14	21	27	16	78		
primary	Joint	23	25	11	15	74		
decision	Decisio							
-maker	n						18.801	0.027
regardin	Doctor	12	14	18	19	63		
g the								
testing?								
Total		74	72	72	67	285		

## 3.14. Chi-square test: Age Group and Reasons for Undergoing Amniocentesis

A chi-square test was conducted to analyse the relationship between maternal age and the primary reason for undergoing amniocentesis(**Table 6**). The test results showed a statistically significant association ( $\chi^2 = 21.68$ , p = 0.041), reinforcing the hypothesis that age influences the motivation behind prenatal genetic testing.

Among participants aged 18–24 years, the most common reason was a doctor's recommendation (27.1%, 16 out of 59), followed by 32.2% citing "other reasons." This trend suggests that younger mothers-to-be may have limited knowledge of genetic risks and rely more on medical advice. Conversely, in the 25–30 years age group, the primary reason was having a previous child with a genetic disorder (36.2%, 25 out of 69), suggesting increased awareness of genetic inheritance risks.

For the 31–35 years age group, 37.7% underwent amniocentesis due to concerns about a prior child's genetic disorder, while 28.3% followed their doctor's recommendation. In the 36–40 years age bracket, "advanced maternal age" emerged as the predominant reason (30.2%), aligning with medical guidelines that classify pregnancies over 35 as high-risk for chromosomal abnormalities. Among women above 40 years, the most cited reason was

physician recommendation (39.2%), reinforcing the role of clinical guidance in genetic screening decisions.

		Reason for	r Undergoi	ng Amnioc	entesis			
			Previous Child	Advan				
		Doctor's	with	ced			Pearso	
		Recomm	Genetic	Matern			n Chi-	р-
		endation	Disorder	al Age	Others	Total	square	value
Age	18 - 24	16	13	11	19	59		
(in	years							
year	25 - 30	13	25	15	16	69		
s)	years							
	31 - 35	15	20	9	9	53		
	years						21.68	0.041
	36 - 40	9	15	16	13	53	21.00	0.041
	years							
	Above 40	20	13	14	4	51		
	years							
Tota		73	86	65	61	285		
1								

## Table 6: Chi-square test

## 3.15. t-test: Patient Group – Awareness and Social Concerns

An independent **t-test** was performed (**Table 7**)to compare the awareness and perception of prenatal genetic testing between pregnant women and their partners. The results revealed a significant difference (t = 2.077, p = 0.039), with pregnant women showing a higher awareness level (M = 1.513, SD = 0.571) compared to their partners (M = 1.363, SD = 0.581). This indicates that women, as primary recipients of prenatal care, are more informed about genetic testing, likely due to direct interaction with healthcare providers.

Similarly, when examining social and ethical concerns, a statistically significant difference was observed (t = 2.133, p = 0.034), where pregnant women (M = 0.626, SD = 0.485) reported greater concerns than their partners (M = 0.495, SD = 0.503). This suggests that women may experience more ethical dilemmas influenced by cultural, religious, and personal beliefs, highlighting the need for couple-based counselling sessions for informed decision-making.

						Std.		
					Std.	Error		p-
Patient group			Ν	Mean	Deviation	Mean	t value	value
Awareness	and	Pregnant	190	1.513	0.571	0.041		
Perception	of	women					2.077	0.039
Prenatal	Genetic	Partners	95	1.363	0.581	0.060	2.077	0.039
Testing								

Table 7: t-test: Patient group - Awareness and Social concerns

				iternational sour	nai oj innovaci	on studies s	(1)(2023)	
Social and Ethic	al Pregnant	190	0.626	0.485	0.035			
Concerns	women					2.133	0.034	
	Partners	95	0.495	0.503	0.052			

## **3.16.ANOVA: Socioeconomic Status and Awareness**

To investigate the variation of awareness and perception of prenatal genetic testing across different socioeconomic groups, a one-way ANOVA was performed (Table 8). As a result of this analysis, it was determined that there was a significant difference between the awareness and perception mean scores according to socioeconomic status (F=3.307; p=0.021). This probably means that knowledge and perception of prenatal genetic screening are influenced by one's economic background. The awareness score did not indicate parity across income groups since it was lower for those whose incomes go above Rs. 50,000 because their mean awareness score read...M = 1.24 and standard deviation was found to be SD = 0.52. For those with an income of Rs. 10,000 - Rs. 25,000, higher awareness was received because their mean awareness scores recorded...M = 1.55 and SD = 0.56. While respondents from the lowest income category, that is below Rs. 10,000, scored a mean of 1.43 (SD = 0.61). On the other hand, those earn between Rs. 25,001 - Rs. 50,000 had a mean of 1.53 (SD = 0.57).

These findings tend toward high awareness and perception in awareness and perception about prenatal genetic testing as a middle-income group. This may be due to a good access of health care resources and information. Strikingly, however, the richest segment shows awareness to be even lower. This may be indicative of their being oriented more to the contact within the medical community concerning information rather than self-illustration or, by their attitude, that genetic testing does not pose any concern. The findings indicated significant differences among income groups, suggesting that financial status plays a role in access to genetic screening information. Further post-hoc analysis is needed to determine specific differences

Awareness and Perception of Prenatal Genetic Testing										
Socio economi c status	N	Mea n	Std. Deviatio n	Std. Erro r	ANOVA					
Less	74	1.43	0.61	0.07						
than Rs.					Awareness and Perception of Prenatal Genetic					
10,000					Testing	Testing				
Rs.10,00	79	1.55	0.56	0.06						
0 –						Sum of				
Rs.25,00						Square		Mean		
0						s	df	Square	F	Sig.
Rs.25,00	87	1.53	0.57	0.06	Betwee	3.235	3	1.078		
1 –					n				3.307	0.021
Rs.50,00					Groups				5.307	0.021
0										

between grou	ips. Table 8: ANOVA:	Socio-economic status an	d Awareness
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		International Journal of Innovation Studies 9 (1) (2025)								
More	45	1.24	0.52	0.08	Within	91.628	281	0.326		
than					Groups					
Rs.50,00										
0										
Total	28	1.46	0.58	0.03	Total	94.863	284		1	
	5									

## **3.17.ANOVA: Education Level & Decision Making & Psychological Impacts Among** Participants

In order for determining influence of education level on the decision-making process and psychological consequences of prenatal genetic testing, one-way ANOVA was conducted (Table 9). The results indicated a statistically significant difference among the groups (F =2.648, p = 0.034), meaning differing educational backgrounds characteristically exhibit differences in psychological impact and involvement in decision-making. The mean scores across various education levels display notable disparities. Participants with undergraduate degrees obtained the highest mean score (M = 2.20, SD = 0.75), pointing to a relatively higher psychological burden accompanied by decision-making involvement. Comparably, those with only high school education (M = 2.09; SD = 0.76) and those with no formal education (M =2.06; SD = 0.66) reported relatively high psychological detriment. In contrast, individuals with postgraduate education had the lowest mean score (M = 1.83, SD = 0.67), indicative of comparatively lower psychological stress and a more systematic approach to decision-making. The findings indicate that education can be crucial to fostering psychological resilience and navigating complex medical decisions. Higher education levels may empower people to better access medical information, improving their critical thinking abilities and, therefore, their confidence in medical decision-making. In comparison, lower education might exacerbate uncertainty and stress due to the limited understanding of and information about the medical procedures and possible outcomes.

Decision-Making Process and Psychological Impact											
			Std.	Std.							
Education		Mea	Deviati	Err							
level	N	n	on	or	ANOVA						
No formal	44	2.06	0.66	0.1							
education				0							
Primary	57	1.90	0.69	0.0	Decision-Making Process and Psychological						
school				9	Impact						
High school	64	2.09	0.76	0.0	Sum						
				9	of Mean						
					Squar Squa						
					es df re F Sig.						

Undergradu	59	2.20	0.75	0.1	Betwe	5.337	4	1.334					
ate degree				0	en								
					Group								
					s				2.64	0.03			
Postgraduat	61	1.83	0.67	0.0	Within	141.1	280	0.504	2.04	4			
e degree				9	Group	07			0	4			
					s								
Total	28	2.01	0.72	0.0	Total	146.4	284						
	5			4		44							

#### 4.DISCUSSION

In our centre, AF testsample utilized for determining abnormal screening outcomes showed similar findings with the studies carried out in other tertiary healthcare institutions in India (Bansal & Jhaveri, 2022). The findings from the retrospective analysis shed light on several crucial aspects of prenatal testing for genetic abnormalities in the Indian context. The study encompassed a diverse cohort of maternal women and their partners, offering a comprehensive understanding of the social impacts and concerns associated with genetic testing. 190 participants were exhibiting diversity in terms of age, educational qualification, occupation, and regional distribution. Most attendees were aged between 25 and 35, with a variety of educational backgrounds and jobs. This mix is important for understanding how genetics research affects society. Gestational testing mainly collects amniotic fluid in the second trimester (16 to 18 weeks), which lowers risks linked to early interventions(Sharma, &Kaul, 2023). The study found a higher rate of chromosomal issues, partly due to the small sample size; a larger group might show lower rates of trisomy. Some important findings pointed to complex cases that need deeper investigation to understand the interacting factors. While most participants felt reassured, the reduced risk of complications is comforting only when paired with a complete understanding and ongoing support(Rose et al., 2020)

Although most amniotic fluid samples showed no chromosomal abnormalities, indicating a reassuring level of genetic stability, the study also identified a range of unusual conditions with different effects. Irregularities like Edward's syndrome, Down syndrome, Patau syndrome, Triploidy XXY, and Neural Tube Defects highlight the important diversity of genetic variations in the studied group (Sadlecki et al., 2018). This reinforces the value of amniocentesis as a key tool that provides essential information for making important medical decisions during pregnancy. Most participants were quite familiar with various prenatal screening methods, especially amniocentesis and maternal tests. Amniocentesis stood out as a well-known method for its accuracy, as many participants noted, which is promising for informed consent regarding genetic screening(Kashyap et al., 2016; Verma et al., 2019).

Majority of the expectant parents have expressed desire for prenatal screening. Recommendations from personal beliefs, healthcare providers, and information access about tests play pivotal roles in decision-making processes (Diadori, 2017). This showed need for a thorough patient education for aiding couples and individuals making informed choices. From an India context, responses on prenatal genetic screening reflectblend of deep-rooted cultural values and modern medical practices. By employing invasive techniques in prenatal screening

poses to be essential for better knowledge and understanding of genetic disorders (Minear et al., 2015). Whilst certain individuals however accept genetic testing as a means for making informed decisions, others tend to view it as challenge for traditional cultural norms. Therefore, incorporation of invasive techniques on prenatal screening holds to be vital in understanding genetic complications and challenges that can likely be encountered by expectant mothers. Comprehensive patient education holds to be crucial such that the couples and individuals confidently navigating complexities of modern medical examination and also whilst respecting the cultural beliefs as well (Seavilleklein, 2009).

The discussions of societal norms and ethics governing prenatal testing in India has carefully balanced cultural traditions, medical advancements, and personal choices(Ghai, &Johri, 2008). India has been constantly navigating between modernity and heritage, decisions revolving around prenatal testing, which includes invasive approaches such as amniocentesis, showcasing shared hopes and values for more inclusive communities (Carlson,& Vora, 2017). The choices that are made during pregnancy, particularly influenced by genetic tests, are deeply emotional as well as personal. The decisions not only affect individual families but also holds a broader implication across the society as well. The study also has uncovered some of the notable insights on the societal impacts of emerging ethical issues and decisions.

Concerns revolving resource allocation, pressure towards terminating pregnancies, concerns revolving disability supports, and potential decrease in the prevalence of such genetic disorders impacted population poses to be the significant aim of prenatal testing. Statistically significant differences between maternal women and their partners in specific concerns, particularly "Pressure to Terminate" and "Impact on Disability Support," indicate varying perspectives within the expectant parent dyad. Expectant parents, guided by traditional family structures and values, navigate decisions influenced not only by medical factors but also by familial expectations and societal norms (Kelley & Rubens, 2010). Also, the study aligns with existing literature highlighting the ethical and societal dimensions of genetic testing. Similar concerns about pressure to terminate pregnancies and concerns with taking testweredocumented in previous studies (Rose et al., 2022; Ravitsky et al., 2021;Poon et al., 2021). The nuanced analysis of regional variations adds depth to the understanding of how geographic factors may influence the social impact of genetic abnormalities in prenatal testing, contributing a unique perspective to the literature.

## **5.CONCLUSION**

The prevalence of genetic abnormalities, as identified via testing AFsamplerevealed a majority of normal fetal development, yet underscored the diagnostic significance of the procedure with notable occurrences of trisomy cases and neural tube defects. This paper addresses the social and psychological impact of prenatal genetic tests on pregnant women and their partners in India. Findings asserted that an awareness of prenatal genetic testing is significantly associated with the level of education, suggesting that individuals with a higher level of education show more awareness. The decision-making pressure differed if the main decision-makers were the couple in that joint decisions tended to have lower levels of stress. Age was the main determinant for deciding whether to go for an amniocentesis; advanced maternal age and genetic conditions in previous offspring made significant contributions to the decision-making process. The study ultimately stresses thenecessitation to impart specific educational intervention approach and counselling support for expectant parents, particularly those with

less education and less income, around prenatal genetic testing, so that they may make informed decisions while undergoing minimum stress. Also, the healthcare providers'influence and their consultancies in general emerge as a pivotal influence in the decision-making process, emphasizing the crucial role of medical professionals in guiding expectant parents. The findings emphasize the need for comprehensive patient education, healthcare provider involvement, and societal awareness to ensure informed decision-making and support for individuals and families navigating the intricacies of genetic abnormalities during pregnancy.

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#### Discussion

The administration of A $\beta_{1-42}$ -oligomer has shown significant (p < 0.05) induction of AD which was reflected in the neurobehavioral and neurochemical changes.Data analysis of the present study showed that GE exhibited a potential ameliorative effect against the A $\beta_{1-42}$  oligomer-associated ADchanges and restored the behavioral and neurochemical changes close to the normal level. These indicates a potential ameliorative effect of GE against the A $\beta_{1-42}$  oligomer-associated AD. Previous research reports stated the neurotoxic effect of  $A\beta_{1-42}$  oligomer in gradual enhancing of the  $\beta$ -amyloid deposition and tau protein accumulation. Moreover, it also enhances the formation of senile plaque in the brain regions of the hippocampus and cortex with cognitive dysfunction and neuronal death, which leads to the progression of mild to severe AD [20,21]. Further studies state that  $A\beta_{1-42}$ accumulation also producesoxidative stress and promotes microglial activation [22,23]. Inflammatory mediators and the accumulation of free radicals lead to the neurodegenerative process [24]. Behavioral assessment from NORT showed that GE group animals exhibited a significant recognition ability of novel objects in NORT in comparison with the AD group and the effects were similar to the effect of reference drug *i.e.*, donepezil treatment, indicating that GE rescued the A $\beta_{1-42}$ oligomer-induced memory loss and cognitive dysfunctions. Our study result lies in parallel with a few other research reports that state the memory rescuing potential of GE [25,26]. Though a large volume of data is available regarding the anti-tumor mechanism of EGFR inhibitor -GE, very seldom its effect on memory and neurodegeneration has been studied. Wang et al conducted a study to determine the molecular mechanisms underlying the pharmacological and genetic effects of EGFR in Aβ- induced memory loss, they assayed the EGFR activation level in the hippocampus region of double transgenic mice through Western blotting. Observations showed that the activated form of phosphorylated EGFR(p-EGFR) level was significantly increased in the mice hippocampus. After 18 days of treatment with GE, the increased p-EGFR level was brought back to a similar level to that of the control group mice, this showed that elevated EGFR activity is well correlated with the Aβ-induced memory loss. Immunoprecipitation studies showed that both A $\beta$ 42 monomers and oligomers were pulled down with wild-type EGFR (EGFRwt). The results obtained from this mechanism-guided study support the hypothesis that EGFR functions as a cell membrane receptor of A<sup>β</sup> peptides, also the A $\beta$  oligometric nuclear activation of EGFR plays a crucial role in leading to memory loss [26]. Moreover, the administration of  $A\beta_{1-42}$  oligomers induced potential alteration of the neurochemical *i.e.*, raised NSE levels in the hippocampus, cerebral cortex, and cerebellum of mice brain samples. NSE is expressed in central and peripheral neurons and also in neuroendocrine cells, which can exist as either  $\gamma\gamma$  or  $\alpha\gamma$  dimeric isozymes. The  $\gamma\gamma$  form of NSE is predominant in neurons, whereas the supporting glial cells such as microglia, oligodendrocytes, and astrocytes express both the  $\alpha\gamma$  form of NSE and non- neuronal enolase (NNE,  $\alpha$ -enolase) [27,28]. Early studies suggested that NSE could be a more potent biomarker for assessing and evaluating neuronal damage and the prognosis of brain injury and brain lesions [29-31]. Previous studies conducted on the investigations of NSE with relevance to AD, as revealed inconsistent findings withfew studies stating elevated NSE levels as

a biomarker for AD [32,33] and some studies stating severity-dependent levels [34], also few other studies stating unaltered levels of NSE [35,36], or even decreased levels of NSE [37]. In the present study, we estimated the NSE level from the brain homogenate of animals in all the groups. AD group mice's brains exhibited an increase in the level of NSE when compared with the normal control group mice; these results were similar to the previous study results [32,33]. However, the administration of GE (2 and 4 mg/kg; *p.o.*) significantly ameliorated this A $\beta_{1-42}$ oligomer-induced neurotoxicity by restoring to the normal level of NSE and the effects were similar to the reference drug DP (2 mg/kg) treated group. Few studies conducted on the antioxidant potential of GE have shown properties like DPPH(2,2dipheny 1-1-picrylhydrazyl) radical scavenging and hydroxyl radical scavenging[38]. This antioxidant property can be a factor in bringing back the neurochemical NSE close to the normal level.

## Conclusion

The administration of EGFR inhibitor *i.e.*, GE has shown to ameliorate the  $A\beta_{1-42}$  oligomer-induced neurotoxicity due to its potent inhibition factivated EGFR, antioxidant, and anti-lipid peroxidative effect. Therefore GE can be a novel synthetic medicine for the management of  $A\beta_{1-42}$ -induced neurodegeneration like AD and other kinds of dementia.

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