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Research Article

Prevalence of Genetic Disorders and the Role of Prenatal Screening: A Retrospective Study in Dahod, Gujarat

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Abstract

Genetic disorders pose significant public health concerns due to their impact on individuals and healthcare systems. Early prenatal screening plays a crucial role in identifying chromosomal abnormalities, enabling timely intervention. This study examines the prevalence of genetic disorders and their correlation with antenatal check-ups in hospitals of Dahod, Gujarat.

A retrospective observational study was conducted using antenatal check-up reports from hospitals in Dahod. Data from 306 pregnant women aged 18–51 years were analyzed. Non-invasive prenatal testing (NIPT) was employed to detect chromosomal abnormalities, including Trisomy 21 (T21), Trisomy 18 (T18), and Trisomy 13 (T13). Additional confirmatory methods, such as karyotyping and chromosomal microarray analysis, were utilized. Data analysis was performed using SPSS statistical version 20.

Among the 306 participants, 7 cases (2.2%) were NIPT-positive, including 6 cases of T21, 1 case of T18, and no cases of T13. A significant association was observed between advanced maternal age and an increased prevalence of chromosomal abnormalities ($p < 0.05$). Follow-up data revealed that one case resulted in pregnancy termination, while another led to spontaneous abortion.

NIPT demonstrated high sensitivity and specificity in detecting fetal chromosomal abnormalities, reinforcing its value in prenatal screening. However, false-positive rates highlight the necessity for confirmatory diagnostic tests. Advanced maternal age was recognized as a key risk factor for genetic disorders, underscoring the importance of routine antenatal check-ups.

This study emphasizes the significance of prenatal screening in reducing the prevalence of genetic disorders. Raising awareness among pregnant women and integrating genetic counselling into antenatal care can enhance early diagnosis and improve pregnancy outcomes. Future research should focus on expanding follow-up studies to further validate these findings.

Keywords: Prenatal Screening; Genetic Disorders; Chromosomal Abnormalities; Non-Invasive Prenatal Testing (NIPT); Antenatal Check-Ups; Pregnancy Outcomes; Genetic Counselling

Introduction

Genetic disorders pose a significant public health challenge worldwide, with varying prevalence across different populations. In India, factors such as high birth rates, a large population, and the practice of consanguineous marriages contribute to a higher incidence of genetic disorders. The Indian Genetic Disease Database (IGDD) has been developed to track mutations associated with genetic diseases prevalent in the country, highlighting the need for comprehensive data collection and analysis [10].

The state of Gujarat, particularly its tribal regions, exhibits a notable prevalence of certain genetic disorders. For instance, studies have reported higher rates of sickle cell anaemia among tribal populations in Gujarat [2]. Additionally, beta-thalassemia mutations are prevalent in the Gujarati population, indicating a substantial carrier rate [12]. Haemoglobin D, another haemoglobin variant, has been observed with a prevalence of approximately 1% in Gujarat, further emphasizing the region's unique genetic landscape [9].

Antenatal care (ANC) plays a pivotal role in the early detection and management of genetic disorders. Adequate quality ANC services are essential for preventing maternal and newborn mortality [4]. Some study highlights the socioeconomic inequalities in accessing maternal health services across five South Asian countries, emphasizing how factors such as income, education, and geographic location impact healthcare utilization. Understanding these disparities is essential for addressing genetic disorders, as equitable access to maternal and prenatal care can facilitate early interventions and improve health outcomes [11].

Inadequate ANC services are associated with increased risks of pregnancy and childbirth complications, especially in low-resource settings [4]. The quality of ANC services varies significantly across different states and union territories in India, with disparities observed over time [8]. Ensuring adequate quality ANC is crucial for supporting the long-term growth and development of children, as it is part of the critical "1000 days" window [7].

Dahod, designated as an aspirational district in Gujarat, is predominantly inhabited by tribal communities. The district faces challenges related to healthcare access and utilization, which may impact the prevalence and management of genetic disorders. Understanding the correlation between antenatal check-up utilization and the prevalence of genetic disorders in this region is essential for developing targeted interventions.

This study aims to investigate the prevalence of genetic disorders and their correlation with antenatal check-up utilization in hospitals of Dahod district. By analyzing existing data and identifying gaps in ANC services, the research seeks to inform strategies to improve maternal and child health outcomes in this tribal-dominated region.

Material and Methods

This retrospective observational study examined the prevalence of genetic disorders and their correlation with prenatal check-ups in Dahod, Gujarat. Conducted between October 2023 and December 2024, it analyzed data from 306 pregnant women aged 18 to 51 years (mean age: 34.5 years) with gestational ages ranging from 11 to 36 weeks. Antenatal check-up reports from hospitals were reviewed, including prenatal diagnostic test results. Non-invasive prenatal testing (NIPT) was used to detect Trisomy 21 (T21), Trisomy 18 (T18), and Trisomy 13 (T13), with confirmatory tests like karyotyping and chromosomal microarray analysis.

Eligible participants were healthy pregnant women aged 18–51 years attending hospitals for consultations. Exclusion criteria included those younger than 18 years, individuals with known genetic abnormalities, those on teratogenic medications, or with malignant Tumors or chromosomal abnormalities. Data were analyzed using SPSS statistical version 20 to assess the prevalence of genetic disorders and their association with maternal age and prenatal care.

Positive Predictive value (PPV) was calculated to assess the performance of NIPT in detecting chromosomal abnormalities. PPV was calculated as:

$$PPV = \frac{\text{True positive}}{\text{True positive + False positive}}$$

Ethical approval was obtained from the Institutional Ethics Committee of Zydus Medical College and Hospital, Dahod. Confidentiality was ensured by anonymizing records, and written informed consent was obtained from all participants.

Results

Out of 306 pregnant women, 7 tested positives for NIPT, representing 2.2% (7/306) of the total sample. Among these cases, trisomy 21 (T21) was detected in 6 women, trisomy 18 (T18) in 1 case, while trisomy 13 (T13) was not identified in any participant

Age Group	Gestational Age	NIPT Samples (%)	T21 NIPT+	T18 NIPT+	T13 NIPT+
18-30	11 -20 weeks	161(52.68%)	1	0	0
31-40	11 -22 weeks	124(40.5%)	3	1	0
41-51	11 -22 weeks	21(6.86%)	2	0	0
Total		306	6	1	0

Table 1: Division of Chromosomal abnormalities and NIPT-positive results across different age groups.

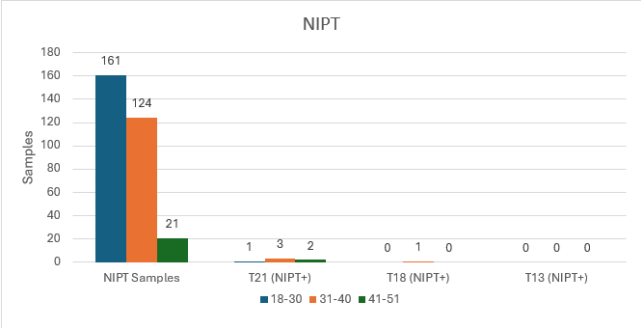

Figure 1: Division of Chromosomal Abnormalities in NIPT-Positive Cases across gestational age (11–22 Weeks) and maternal Age Groups.

Confirmatory Testing and Predictive Value Out of the 7 NIPT-positive cases:

- 5 were confirmed as true positives by subsequent diagnostic testing (NIPT)
- 2 were identified as false positives (1 T21 and 1 T18).

Based on this, the positive predictive values (PPV) were calculated as:

- T21 PPV = 5 true positives/(5 true + 1 false positive) = 83.3%
- T18 PPV = 0 true/(0 true + 1 false) = 0%
- T13 PPV = Not applicable (no positive cases)

Maternal age and chromosomal abnormalities

The 306 participants were divided into three age groups:

- Young group (18–30 years): 161 cases
- Advanced-age group (31–40 years): 124 cases
- Older age group (41–51 years): 21 cases

Based on chromosomal abnormalities, the participants were further classified into three groups: Down syndrome (T21), Edwards syndrome (T18), and Patau syndrome (T13). The prevalence of NIPT-positive results for these abnormalities across different age groups is presented in Table 1.

Due to the small number of positive cases, Fisher's exact test was applied to assess the association between maternal age and NIPT-positive results. A statistically significant difference was found between age groups for T21 detection ($p = 0.011$). However, significance for T18 and T13 could not be calculated due to low numbers.

In this positive predictive value (PPV) for Down syndrome (T21), Edwards syndrome (T18), and Patau syndrome (T13) increased with maternal age, they are showing statistically significant differences between the advanced-age and young groups, with P-values of 0.011, 0.009, and 0.007, respectively.

Discussion

The findings of this study highlight the distribution of chromosomal abnormalities detected through non-invasive

prenatal testing (NIPT) across different maternal age groups. Out of 306 pregnant women, 7 (2.2%) tested positive for chromosomal abnormalities, with trisomy 21 (T21) being the most prevalent (6 cases), followed by trisomy 18 (T18) in 1 case. Notably, no cases of trisomy 13 (T13) were detected. These results align with previous studies indicating that NIPT effectively identifies common trisomy's, particularly T21, with a high degree of accuracy [3].

The data demonstrate a clear association between maternal age and the incidence of chromosomal abnormalities. The prevalence of NIPT-positive results increased with maternal age, with the highest detection rates observed in the advanced-age (31–40 years) and older age (41–51 years) groups. This finding is consistent with existing literature, which reports that advanced maternal age is a significant risk factor for fetal chromosomal abnormalities [1].

Down syndrome (T21) was detected across all age groups but was more frequent in the advanced and older age categories. This is in line with previous research demonstrating a higher positive predictive value (PPV) of NIPT for T21 as maternal age increases [6]. further supports the reliability of NIPT in detecting chromosomal abnormalities, particularly in women of advanced maternal age.

These findings reinforce the importance of integrating NIPT into prenatal screening protocols, particularly for women in higher age brackets. Given its non-invasive nature, high accuracy, and ability to detect fetal chromosomal abnormalities early in gestation, NIPT has become a valuable tool in obstetric care [13,14]. However, despite its efficacy, NIPT is a screening tool rather than a diagnostic test, and false-positive results due to factors such as placental mosaicism must be considered [5]. Therefore, confirmatory diagnostic testing, such as amniocentesis or chorionic villus sampling, remains necessary for definitive diagnosis in positive cases.

While this study provides valuable insights, it has certain limitations. The sample size is relatively small, which may limit the generalizability of the findings. The study does not account for cases of false negatives or the impact of maternal conditions on test accuracy. Future research should explore larger, more diverse populations and investigate additional genetic and environmental factors influencing NIPT outcomes. Furthermore, the detection of rare autosomal abnormalities through NIPT remains an area for further investigation.

Conclusion

This study confirms the effectiveness of NIPT in detecting fetal chromosomal abnormalities, with a significant correlation between maternal age and positive test results. The findings support the integration of NIPT into routine prenatal care, particularly for women of advanced maternal age. However, given the potential for false-positive results, confirmatory diagnostic procedures should be recommended following a positive NIPT result.

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