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THE PREVALENCE OF CHROMOSOMAL ABNORMITIES DIAGNOSED PRENATALLY IN A AN AND POPULATION

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ABSTRACT: Background: The most important factor contributing to human genetic problems is chromosomal abnormalities. It is crucial to choose screening tests and diagnostic procedures that are exact, accurate, safe, and able to be conducted during early pregnancy in order to make the best-informed choice possible while taking into account the probable outcomes of pregnancy. **Method and Material:** In the present prospective study chromosomal analysis was done for various types of suspected 900 referred patients. The patients were referred from mainly Gynaecology hospital Usha Nursing Home and Sat Kaival Hospital, Anand in association with Gene Care Accuris Laboratory, Surat from June 2021 to March 2023. **Result:** The overall frequency of chromosomal abnormalities was 2.5% (23/900). Out of 23 cytogenetic abnormal patients, numerical abnormalities were found in 21(2.33%) and structural abnormalities we detected in 02(0.22%) patients. The most common autosomal abnormalities were Down' syndrome 18(2%). Another abnormality was Edward's Syndrome 03(0.33%). Chromosomal structural disorder occupied 02(0.22%) including robertsonian translocation. **Conclusion:** To avoid the delivery of foetuses with chromosomal disorders, karyotype screening of amniotic fluid is a crucial strategy. Our results underline the significance of cytogenetic investigations in individuals with signs of prenatal diagnosis because an aberrant finding not only gives patients the option of terminating or continuing their pregnancies but also serves as a foundation for genetic counselling and helps in creating a healthier society.

INTRODUCTION: The most important factor contributing to human genetic problems is chromosomal abnormalities. Many kinds of chromosomal abnormalities, both structural and numerical, have been clinically suspected of having genetic abnormalities.

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To identify chromosomal deletion, translocation, duplication, inversion, and aneuploidy of the autosomes and sex chromosomes, cytogenetic investigation is crucial [1].

Other issues include the absence of treatment options for chromosomal disorders and false-positive screening tests in sonography and/or maternal serum. It is crucial to choose screening tests and diagnostic procedures that are exact, accurate, safe, and able to be conducted during early pregnancy in order to make the best-informed choice possible while taking into account the probable outcomes of pregnancy in between 2%

and 5% of all live births, genetic and congenital abnormalities are reported to occur by the World Health Organisation. In developing nations, these changes are responsible for 50% of all childhood fatalities. Additionally, they are mostly to blame for prenatal and neonatal mortality in underdeveloped nations [2]. Cytogenetics analysis has grown significantly in importance over the past ten years as a tool for genetic counselling, which deals with the human issues connected to the presence or risk of a genetic disorder in a family and aids in understanding the diagnosis, prognosis, and available management, as well as the genetic basis, likelihood of recurrence, and available options [3]. Objective of the present study is to assess the prevalence and kind of chromosomal abnormalities in high-risk pregnancies utilizing karyotype and FISH analysis of amniotic fluid cells.

MATERIAL AND METHODOLOGY: After approval from the institutional ethics committee, a prospective clinical study was conducted at Usha Nursing Home and Sat Kaival Hospital, Anand in association with Gene Care Accuris Laboratory, Surat from June 2021 to March 2023. After diagnostic counselling acquiring family and gestational history. The patients also received comprehensive prenatal diagnosis. Informed written consent was taken. Pregnant women in whom serum screening tests (Double marker & Quadmarker) showed a high fetal risk underwent prenatal testing. Maternal serum screening consisted of tests for the free beta-human chorionic gonadotrophin (free β-hCG) and pregnancy-associated plasma protein A (PAPP-A) in the first trimester, and in the second trimester the Quadmarker test for alpha-fetoprotein (AFP), β-hCG, and unconjugated estriol (uE3) and Inhibin A. PRISCA software was used for prenatal screening and calculated the relative multiples of median (MoM) and risk values. Gynaecologist was responsible for ordering all the investigation. Ultrasound-guided trans- abdominal puncture was the technique used to collect samples of amniotic fluid. In long-term cell cultures using Amniomax medium at 37 °C in a CO_2 incubator, amniotic fluid or any other foetal sample acquired was grown. After 6 days it was checked whether cell was sticked or not. If not sticked incubate for another 2 to 3 more days. After cell were sticked, they were arrested by Colchicine in methaphase. After chromosome harvesting, standard cytogenetics methods were applied to obtain spread chromosomes on the slide. G-Bands were induced by trypsin treatment and a resolution of the least 400 bands were obtained. Minimum 20 methaphase were analyzed for each case and karyotype were obtained.

RESULT: In our study 900 cases were examined, out of 45 prenatal cases had positive biochemical screening results, 33 had positive in Double marker and 12 Qquadruple marker results along with ultra-solography. On the basis of Biochemical screening, all 45 high risk patients referred to the NIPT (Non-invasive) confirmation test.

TABLE 1: DISTRIBUTION OF PATIENT ON THE BASIS OF NIPT TEST RESULT

No of Patient(NIPT Test)= 45	
High Risk	**Low Risk**
30 (66.66%)	15 (33.33%)

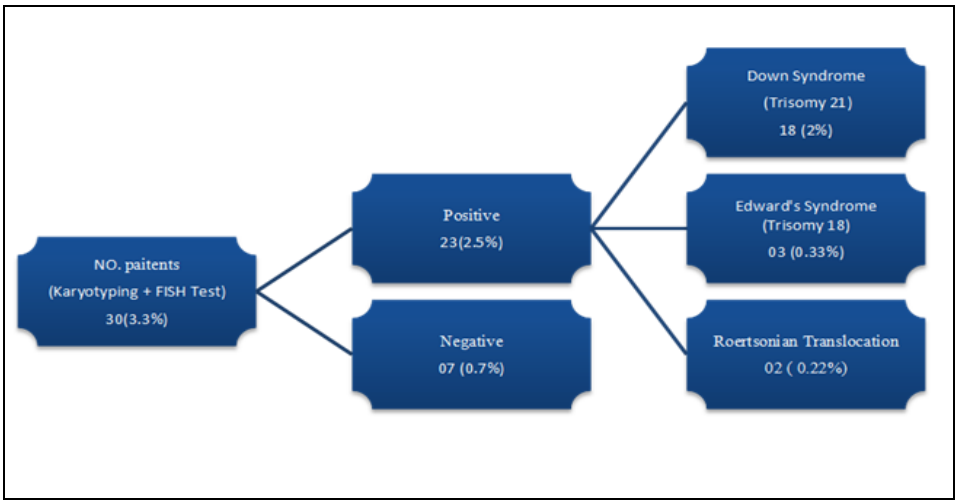


FIG. 1: DISTRIBUTION OF HIGH RISK PATIENTS ON THE BASIS OF RESULT OF KARYOTYPING +FISH TEST

TABLE 2: RESULT OF TRISOMY 21

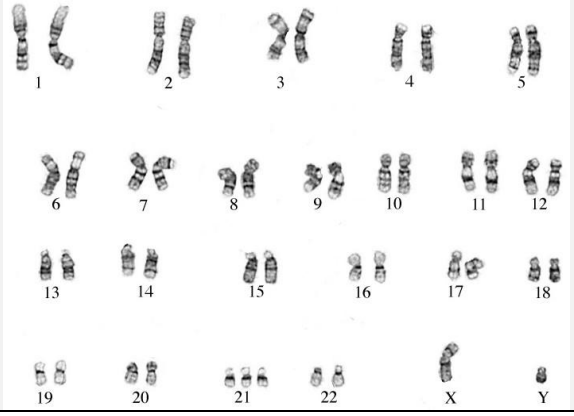
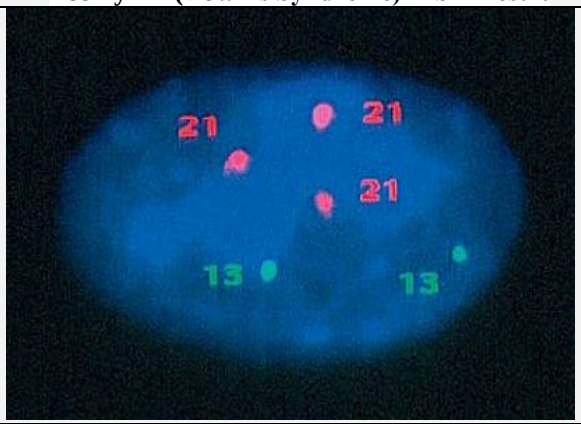
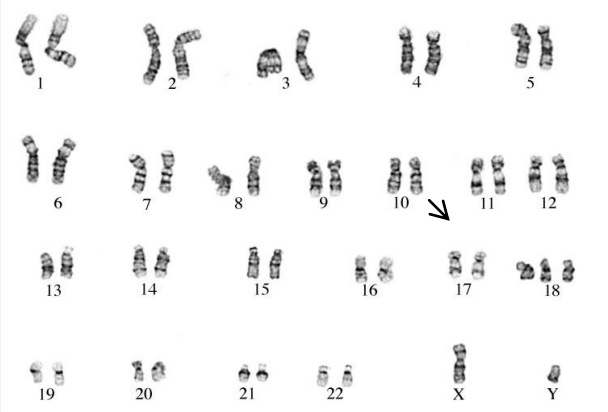
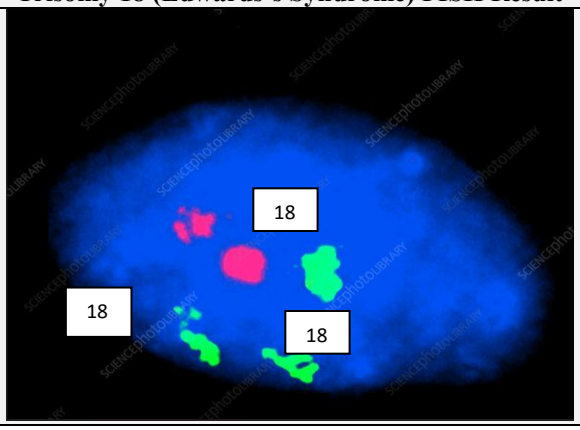
Trisomy 21 (Down's Syndrome) Karytotyping Result	Trisomy 21 (Down's Syndrome) FISH Result
	

TABLE 3: RESULT OF TRISOMY 18

Trisomy 18 (Edwards's Syndrome) Karytotyping Result	Trisomy 18 (Edwards's Syndrome) FISH Result
	

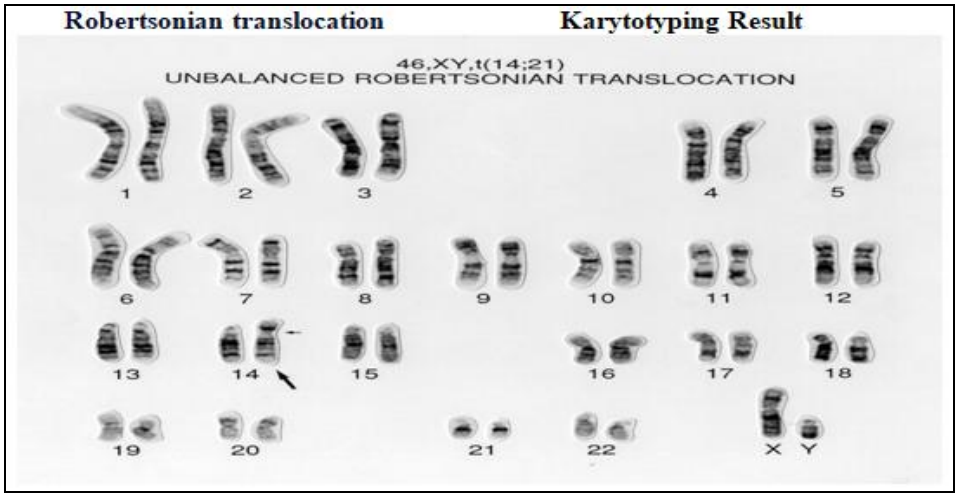


FIG. 2: RESULT OF ROBERTSONIAN TRANSLOCATION

TABLE 4: FREQUENCY OF CHROMOSOMAL ABNORMALITIES WITH RESPECT TO INDICATION

Indication	Types of Chromosomal Abnormalities			
	Total	Trisomy 21, 47, XY/XX,+21	Trisomy 18, 47, XY/XX, +18	Robertsonian Translocation 46, XY, rob (14; 21) (q10; q10+21)
AMA	14	08	06	00
Previous Child History for chromosomal Abnormalities	09	05	04	00
Maternal Serum Testing with Ultrasound (n=23)	23	18	03	01

DISCUSSION: The most frequent genetic illnesses that cause birth malformations in newborns are chromosomal abnormalities. Chromosome abnormalities occur frequently (around 0.5% of live births, 5% to 13% of stillbirths, and 0.5% to 0.5% of neonates. Foetal abnormalities currently have no effective treatments. Second-trimester amniotic fluid cell karyotyping is a crucial preventive tool for prenatal detection and prompt cessation of abnormal pregnancies [4]. After evaluating in our study, we found the incidence and type of chromosomal abnormalities in amniotic fluid samples using conventional cytogenetics analysis. In our study, the invasive procedure was performed between 19 and 25 weeks of gestation Weeks. Similar, to other studies the most frequent indications for amniocentesis were positive maternal serum screening, abnormal ultrasound findings and advanced maternal age. We found numerical abnormalities in 2.33% and structural in 0.22% of cases. So far, the most frequent single abnormality was trisomy 21 (2 %), while other studies done by (Burada F *et. al* 2018) [1] had 3%, (Gu X *et al* 2020) [5] had 4.4%) which is similar to present study while studied done by (Wang W *et al* 2020) [6] had 26%, (Pandey P *et al* 2018) [7] had 93%, (Dai R *et al* 2019) [8] had 70.1%, (Li H *et al* 2019) [8] had 48.32%, and (Zhang S *et al* 2021) had 37.44% showed higher prevalence of Trisomy 21 because this study had a greater proportion of pregnant women and was a retrospective analysis with a longer duration frame. The reported incidence of prenatal chromosomal abnormalities is variable, while some studies indicated similar results with our finding. Another common frequent abnormalities was Trisomy 18 (0.33%), while other study done by (Gu X *et al* 2020) [5] had 1.33%, which is similar to present study while studied done by (Zhang S *et al* 2021) had 11.18%, (Li H *et al* 2019) [10] had 14.25%, (Dai R *et al* 2019) [8] has 16.2%, and (Wang W *et al* 2020) [6] had 24.5% showed higher prevalence of Trisomy 18 because this study had a greater proportion of pregnant women and was a retrospective analysis with a longer duration frame. Next abnormalities were Robertsonian translocation (0.22%). Similar observation was made by (Burada F *et.al* 2018) [1] had 0.44%, (Gu X *et al* 2020) [5] had 0.08% and (Li H *et al* 2019) [10] had 2.6% showed almost similar result. In our study no of pregnant women is low or compared to most of the other studies. Cytogenetic analysis is an essential tool in genetic counselling to establish a definitive diagnosis, to estimate the risk of recurrence of the chromosomal disorders in future pregnancies, and to decide clinical management, which may account for the high prevalence of chromosomal abnormalities despite the availability of advanced prenatal diagnostic techniques in our country [11].

TABLE 5: COMPARISON OF FREQUENCY OF CHROMOSOMAL ABNORMALITIES IN POPULATION OF THE PRESENT STUDY WITH OTHER STUDY

Studies	Total no of patients examined for amniocentesis	Frequency of chromosomal abnormalities found (%)
Present Study- Anand	30	23(2.5%)
Choudhari R [9] (2022) Maharashtra	200	20(10%)
Xie D [10] (2021) China	2883890	3181(0.11%)
Pal A [3] (2020) Maharashtra	2215	271(12.23%)
Dai R [8] (2019)Northeast China	4953	204(4.12%)

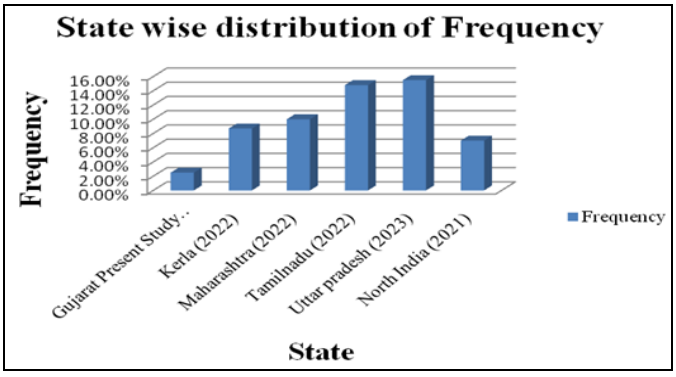


FIG. 3: COMPARISON OF STATE WISE FREQUENCY OF CHROMOSOMAL ABNORMALITIES IN THE PRESENT STUDY WITH OTHER STUDY

CONCLUSION: To avoid the delivery of foetuses with chromosomal disorders, karyotype screening of amniotic fluid is a crucial strategy. Our results underline the significance of cytogenetic investigations in individuals with signs of prenatal diagnosis because an aberrant finding not only gives patients the option of terminating or continuing their pregnancies but also serves as a foundation for genetic counselling and helps in creating a healthier society.

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CONFLICTS OF INTEREST: Nil

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