e-ISSN: 0976-822X, p-ISSN:2961-6042

Available online on http://www.ijcpr.com/

International Journal of Current Pharmaceutical Review and Research 2026; 18(8); 486-505

Original Research Article

Correlation of Karyotyping Pattern in Infants and Children with Congenital Anomalies – A Cross-Sectional Study in a Tertiary Care Centre

Raksha Revanna[1], Chandana A.[2], Ashwini D.P.[3]

[1]Senior Resident, Department of Paediatrics, MIMS, Mandya, Karnataka, India
[2]Assistant Professor, Department of Paediatrics, MIMS, Mandya, Karnataka, India
[3]Junior Resident, Department of Paediatrics, MIMS, Mandya, Karnataka, India

Received: 01-05-2026 / Revised: 15-06-2026 / Accepted: 21-07-2026
Corresponding author: Dr. Raksha Revanna
Conflict of interest: Nil

Abstract

Background: Congenital anomalies are the leading cause of infant mortality. In 40%–60% of cases of malformations, the cause of congenital anomaly is unknown. Genetic factors such as chromosomal abnormalities and mutant genes account for approximately 15% of cases. Chromosomal abnormalities, which may be numerical or structural, are important causes of birth defects Deficiencies in diagnostic capabilities and lack of reliability of medical records and health statistics underestimate the birth prevalence of congenital anomalies in the developing world. The most common serious congenital disorders are congenital heart diseases, neural tube defects, and down syndrome.

Objective: To determine the chromosomal abnormalities (structural and numerical) in congenital anomalies of suspected genetic causes, by Karyotyping.

Methods: A total of 35 cases of congenital anomalies with suspected genetic abnormalities were studied. Peripheral blood from these cases was collected and fifteen–twenty metaphase spread was examined under trinocular research microscope.

Results: In our study, 12 babies had abnormal karyotyping (34.3%) and 23 had normal karyotyping (65.7%). In the study 68.6% had consanguineous marriage and 31.4% had non-consanguineous marriage. Parental consanguinity was significantly associated with an increased risk of congenital anomalies**.** In the study 57.1% of them were males and 42.9% were females. Among Females, 20% had abnormal karyotyping and among males, 53.3% had abnormal karyotyping. There was significant association between Gender and karyotyping. Abnormal karyotyping was common among males with congenital anomalies.

Conclusion: Anomalies were most likely to be in the genitourinary system. Maternal history of parental consanguinity was significantly associated with an increased risk of congenital anomalies**.** There was significant association between Gender and karyotyping. Abnormal karyotyping was common among males with congenital anomalies**.** Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

Keywords: Congenital anomaly, Karyotyping, Downs syndrome, Genetics, Mutation.

DOI: 10.25258/ijcpr.18.8.77

Introduction

The early intrauterine period during 3rd – 8th weeks of gestation is vital for the normal development of organs and organ system or organogenesis [1]. It was observed that better maternal care and improved living standards have minimal effects on the overall frequency of congenital malformations. [2]

A congenital anomaly may be defined in terms of physical structure as a malformation, an abnormality of bodily structure or form usually found at birth or during the first few weeks of life, or defined more widely to include functional disturbance as a defect, any irreversible condition exiting in a child before birth in which there is sufficient deviation in the usual number, size, shape, location or inherent character of any part, organ, cell or cell constituent to warrant its designation as abnormal. [3,4] A congenital anomaly is thus any alteration present at birth of standard anatomic structure and has cosmetic, medical or surgical significance. The birth of an infant with significant malformations, whether diagnosed antenatally or not, evokes an emotional parental response.[5]

According to the World Health Organization document of 1972, congenital malformations should be confined to structural defects at birth. [6] However, as per the more recent WHO fact sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, present at birth. [7] Congenital anomalies are a significant cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. [8,9] It is not only a leading cause of fetal loss but also contributes significantly to preterm birth, childhood and adult morbidity, along with considerable repercussions on the mothers and their families. [10] In 40%–60% of cases of malformations, the cause of the congenital anomaly is not known. Genetic factors such as chromosomal abnormalities and mutant genes account for approximately 15% of cases. Chromosomal abnormalities, which may be numerical or structural, are important causes of congenital disabilities. [11]

The birth prevalence of congenital anomalies in the developing world is underestimated by deficiencies in diagnostic capabilities and the lack of reliability of medical records and health statistics. The most common congenital severe disorders are heart defects, neural tube defects, and Down syndrome. [12] Cytogenetics with Giemsa banded peripheral leukocyte karyotype is the gold standard and should be performed in most evaluations. [13] Congenital anomalies are classified into either single-system or multiple-system malformations [14]. The first type affects a single organ system or body part, and the second affects more than one organ system or body part. Significant congenital anomalies are those that, if uncorrected, could result in considerable impairment of the normal body functions or even reducing life expectancy. [15–17] Minor congenital anomalies cause no disability or have no significant physical or functional effects and can be regarded as normal variants. [15,18] Fetal anomaly scanning is the most effective method of reducing the prevalence of congenital abnormalities and increasing the survival rate of those born with these issues [19].

The finding of a correctable abnormality can serve to indicate that delivery should take place in a centre with paediatric surgery facilities, and the discovery of a severe uncorrectable abnormality might result in offering pregnancy termination. [19]

Survivors of congenital anomalies might have lifelong physical, mental, visual, and auditory disabilities if not managed appropriately [20], which can negatively affect the human and economic life of the person concerned, as well as their families and communities. Parental emotional responses, such as denial, feelings of guilt, worry, grief, and shame, occur after the birth of an infant with significant anomalies.

This response occurs even if the anomalies were diagnosed antenatally, highlighting the significance of proper counselling. [21] It is the most crucial cause of under-five mortality in developing nations like India and accounts for about 61 to 69.9/1000 of all live births. [22] This high prevalence warrants the need to take immediate steps to tackle the problem of a war footing, more so when 70% of these defects are preventable. [22] India ranks second in the world in the reported occurrence of congenital anomalies in neonates and children. [22] This fact highlights the urgency and importance of documenting all congenital malformations occurring in neonates born in hospital settings to focus and develop appropriate preventive and remedial strategies. Quantifying congenital disabilities in a population is a felt need as it helps in the appropriate allocation of the health budget to tackle and reduce perinatal, neonatal and infant mortality rates. [23]

Knowledge about the prevalence of congenital anomalies is useful for obtaining baseline rates, documenting changes over time, and identifying clues to the etiology of conditions. This knowledge is also helpful to plan and assess antenatal screening for congenital anomalies, especially for high-risk populations. [24] The worldwide incidence of congenital disorders is estimated at 3-7%, but actual numbers vary widely between countries. [25] Congenital malformations affect 2.5% of infants at birth and are responsible for about 15% of perinatal mortality in India. [26,27] Approximately 66% of major malformations have no recognized aetiology, and most of them have a multifactorial inheritance. [3] These defects can occur for many reasons including inherited genetic conditions, poor diet, and toxic exposure of the fetus for example, to alcohol, birth injury and, in many other cases, for unknown reasons. [28] The present study might be of help in early and better diagnosis of congenital anomalies, particularly those presenting with chromosomal abnormalities, so that counselling can be started as soon as possible. It might be suggested that Karyotyping of cases of congenital anomalies should be included in their routine investigation. Hence there is a need to study of karyotyping in infants and children with congenital anomalies. Only a few studies are done in India, and in order to broaden our understanding of this subject, this study is undertaken.

Objectives: To determine the chromosomal abnormalities (structural and numerical) in congenital anomalies of suspected genetic causes, by karyotyping.

Materials & Methods

Source of the Data: Study place: Sapthagiri Institute of Medical Sciences and Research Centre, Bengaluru.

Study Subjects: Infants and Children with congenital anomalies admitted in Sapthagiri Institute of Medical Sciences and Research Centre

Study Period: 18 months

Study Design: Cross sectional study

Sampling Technique: Universal sampling

Inclusion Criteria

1. Infants and children with congenital anomalies.
2. Age-< 18 years

Exclusion Criteria

1. Patient not willing to give informed consent for karyotyping

Methodology

A total of 35 cases of congenital anomalies with suspected genetic abnormalities, of both outpatients and inpatients, attending Sapthagiri Institute of Medical Sciences and Research Centre, Bangalore, Karnataka were studied and cases with known teratogenic causes were excluded.

The informed consent was obtained from the parents [Annexure-1] and their data were collected. Full maternal history regarding antenatal history, perinatal history, family history, age, parity, drug intake and radiation exposure during pregnancy was taken. A family pedigree was constructed for all cases. All infants and children were examined to determine age, birth weight, minor or gross external anomalies. Type of abnormality and karyotype of the cases were the outcome variables.

Peripheral blood from these cases was collected and fifteen–twenty metaphase spread was examined under trinocular research microscope. The cultivation of cells (lymphocytes) produced from a specimen is the first step in the karyotype process. Colchicine is administered after period of cell growth and multiplication to arrest cells in metaphase, poisoning the mitotic spindle.

The cells are then exposed to a hypotonic solution, which causes them to explode. The nuclei are then fixed using a chemical fixative, placed on a glass slide, and treated with various dyes to reveal the chromosomes structural properties.

G-banding involves treating metaphase chromosomes with trypsin first, then staining them with Giemsa. Trypsin partially digests the chromosomal proteins, thereby relaxing the chromatin structure and allowing the Giemsa dye to access the DNA, revealing chromosomal structural characteristics. The best metaphase spread was selected, photographed, printed, and karyotypes were prepared. Congenital anomalies were classified using ICD-10 classification. Statistical analysis was performed using t-test, chi-square test to investigate the significances of different variables.

Assessment Tools: ICD-10 classification of congenital anomalies.

Sample Size Calculation: According to Yasir A Mohammed et al, conducted a cross sectional study to document prevalence of congenital malformations detected at birth and also predisposing factors, A prevalence of 2.06% was found. As per this study the prevalence has been taken as 2.06 in our study-

n = 4pq \ d2 P- 2.06%

P- Prevalence q-100-2.06 = 97.94

q- 100-p d- 5

d- Allowable errors

n = 4*2.06*97.94/25 = 32.28 – approx. 35

SAMPLE SIZE: 35

Statistical Analysis

- Data was entered into Microsoft excel data sheet and was analyzed using SPSS 22 version software. Categorical data was represented in the form of Frequencies and proportions.
- MS Excel and MS word were used to obtain bar diagram.
- Chi-square test was used as test of significance for qualitative data. Continuous data was represented as mean and standard deviation.
- p value (Probability that the result is true) of <0.05 was considered as statistically significant after assuming all the rules of statistical tests.
- Statistical software: MS Excel, SPSS version 22 (IBM SPSS Statistics, Somers NY, USA) was used to analyse data.

Results

A total of 35 cases with congenital anomaly were included in this study. Following observations were seen in this present study.

Statistical Analysis: Data was entered into Microsoft excel data sheet and was analyzed using SPSS 22 version software. Categorical data was represented in the form of Frequencies and proportions. Chi-square test was used as test of significance for qualitative data.

Graphical representation of data: MS Excel and MS word was used to obtain various types of graphs such as bar diagram, Pie diagram. p value (Probability that the result is true) of <0.05 was considered as statistically significant after assuming all the rules of statistical tests.

Statistical software: MS Excel, SPSS version 22 (IBM SPSS Statistics, Somers NY, USA) was used to analyse data.

Results:

Table 1: Age Distribution of subjects

		Count	%
Age	Below 1 Month	13	37.1%
	1 Month to 1 year	8	22.9%
	1 Year to 5 Years	7	20.0%
	5 to 10 years	3	8.6%
	>10 years	4	11.4%
	Total	35	100.0%

In the study majority of Children were in the age group <1 Month (37.1%), followed by 1 Month to 1 year (22.9%), 1 Year to 5 Years (20%), 5 to 10 years (8.6%) and >10 years (11.4%).

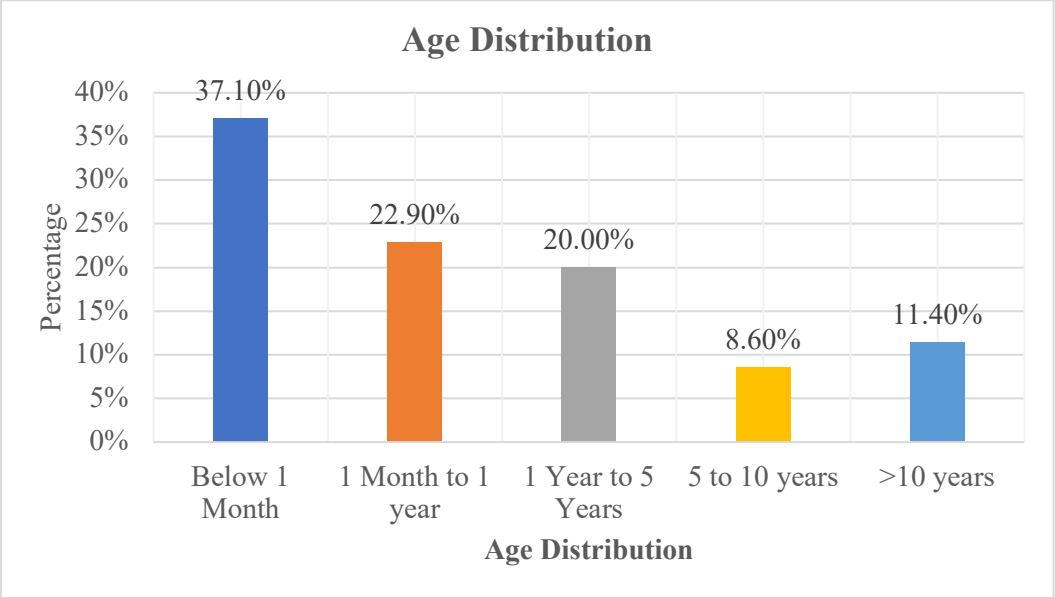

Figure 1: Bar diagram showing Age Distribution

Table 2: Consanguinity distribution

Consanguinity	N = 35	%
Non-Consanguineous marriage	11	31.4%
Consanguineous marriage	24	68.6%

In the study 68.6% had consanguineous marriage and 31.4% had non-consanguineous marriage.

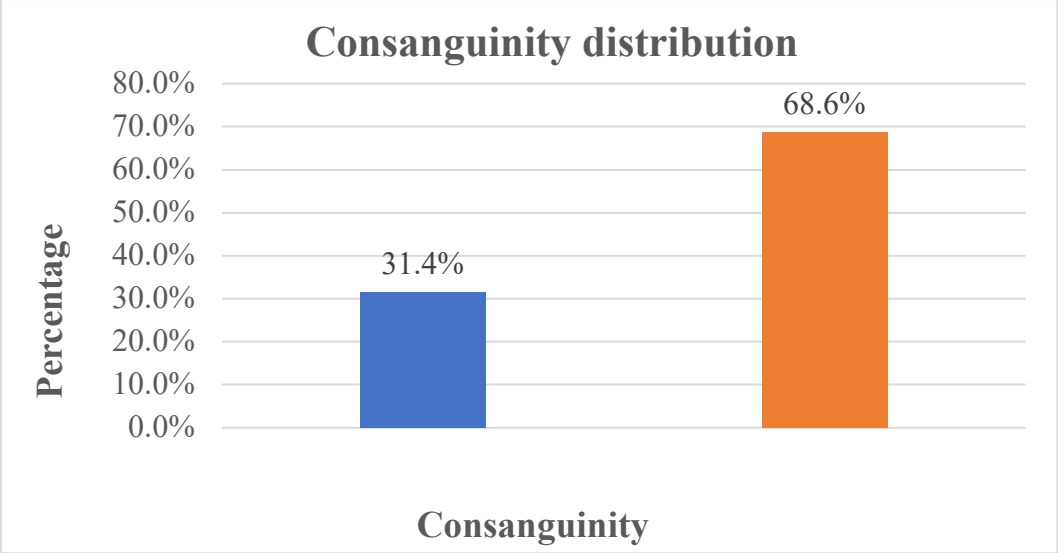

Figure 2: Bar diagram showing Consanguinity distribution

Table 3: Gestational age of the child

Gestational age	N=35	%
Preterm	3	8.6%
Term	32	91.4%

In the study 91.4% were term pregnancy and 8.6% were preterm pregnancy.

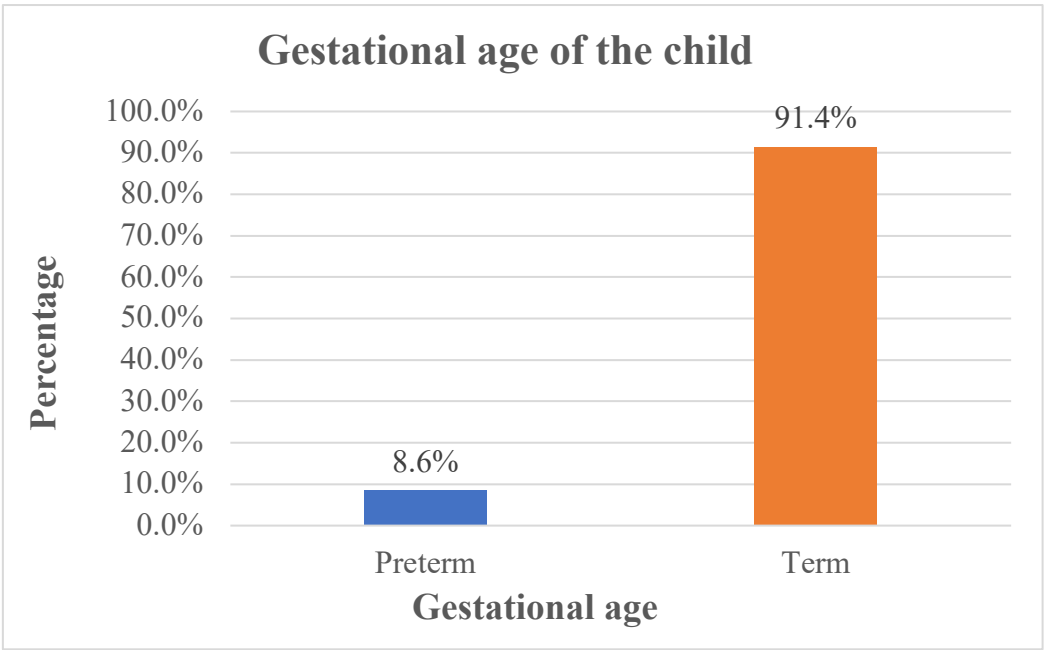

Figure 3: Bar diagram showing Gestational age of the child

Table 4: Gender of the child

Gender	N=35	%
Male	20	57.1%
Female	15	42.9%
Total	35	100.0%

In the study 57.1% of infants were males and 42.9% were females.

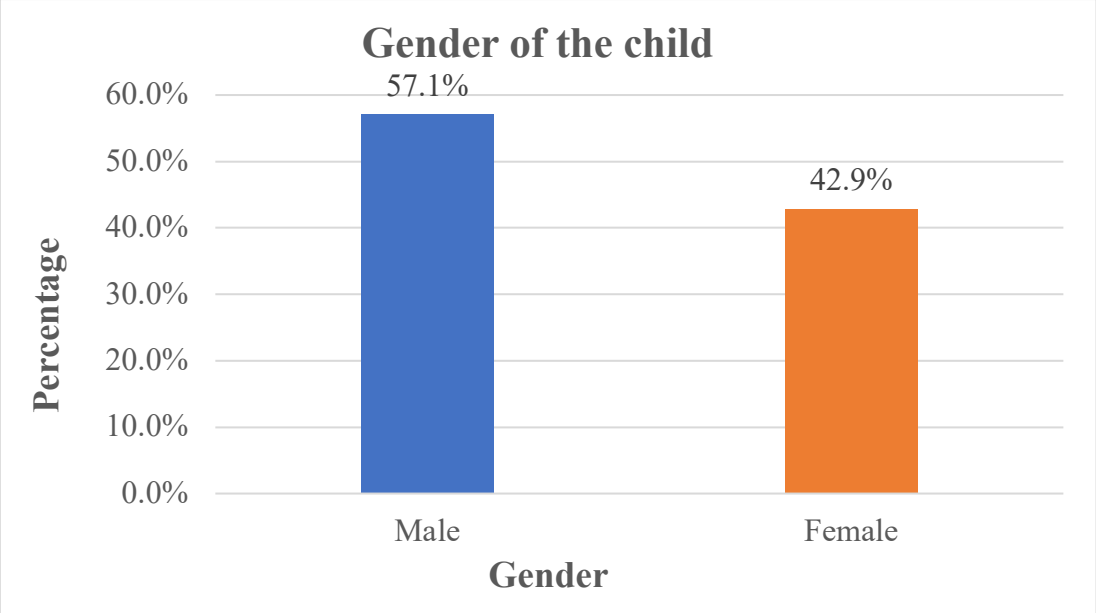

Figure 4: Bar diagram showing Gender of the child

Table 5: Birth Weight distribution

		Count	Column N %
Birth Weight	<2.5 Kg	12	34.3%
	>2.5 Kg	23	65.7%
	Total	35	100.0%

In the study 34.3% of children had Low Birth weight and 65.7% had Normal Birth weight. Mean Birth weight was 2.6886 ± 0.493 Kg.

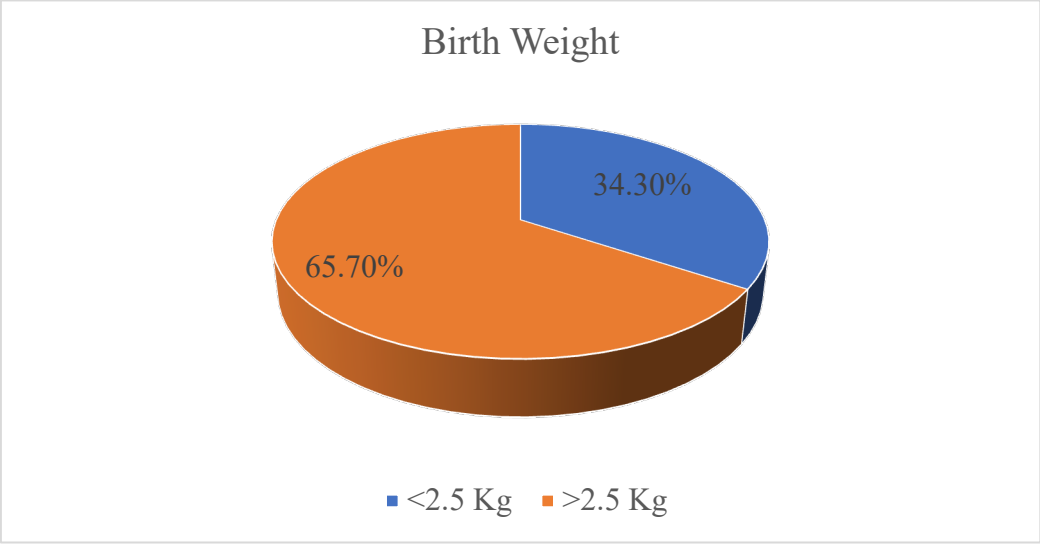

Figure 5: Pie diagram showing Birth Weight Distribution

Table 6: H/O Congenital Anomaly distribution

H/O Congenital Anomaly	N=35	%
Yes	1	2.9%
No	34	97.1%
Total	35	100.0%

In the study 2.9% had family H/O Congenital Anomaly.

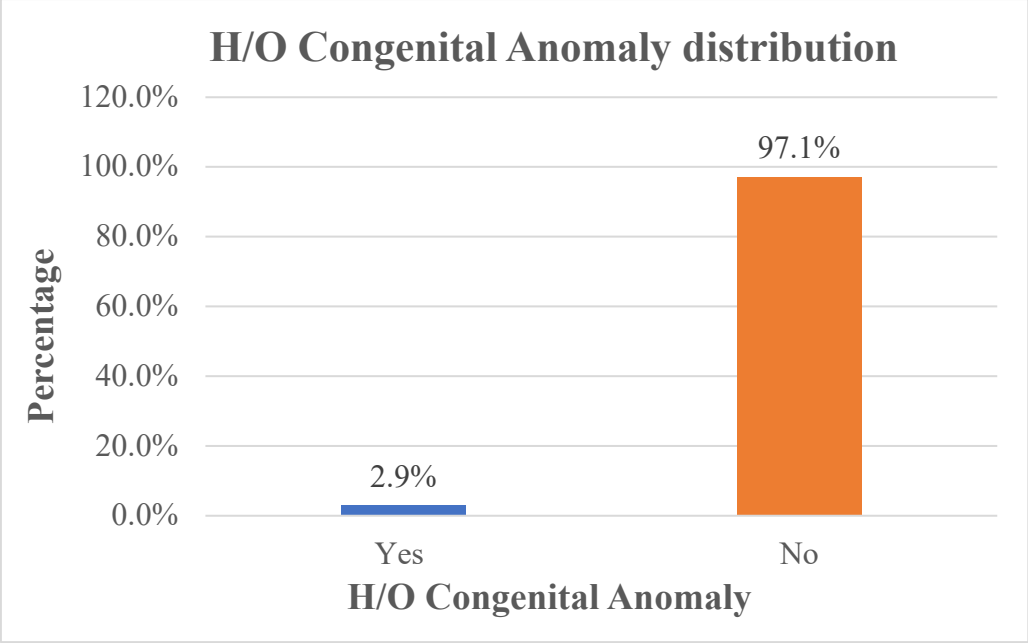

Figure 6: Bar diagram showing H/O Congenital Anomaly distribution

Table 7: Maternal Drug Intake distribution

Maternal Drug Intake	N=35	%
Yes	7	20.0%
No	28	80.0%
Total	35	100.0%

In the study 20% of subjects had history of Maternal Drug Intake.

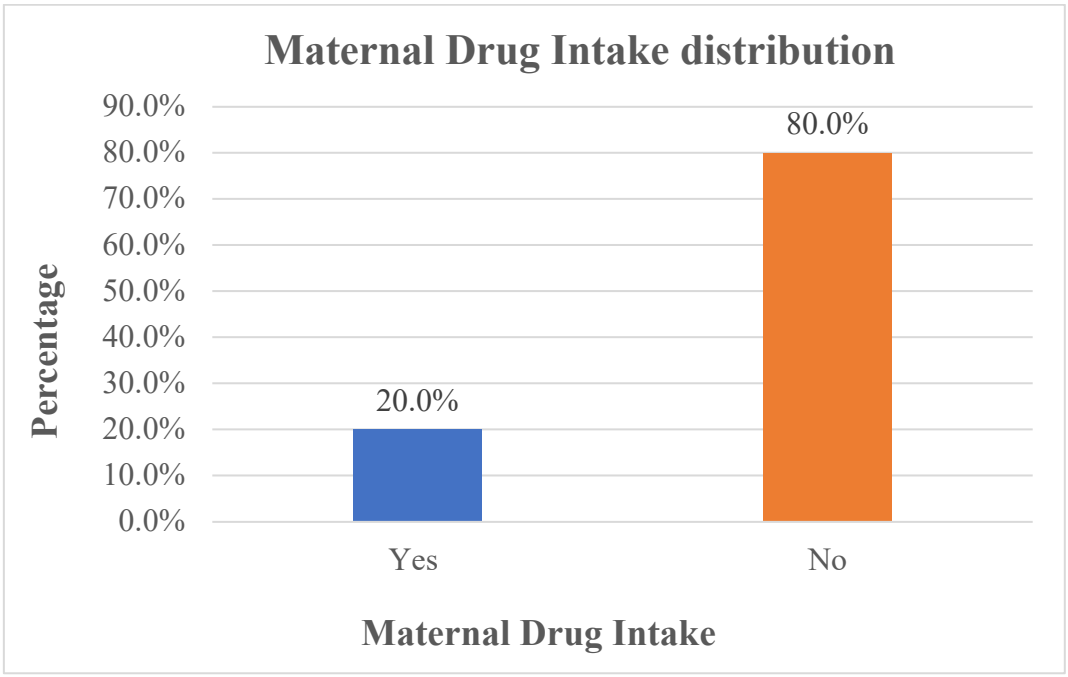

Figure 7: Bar diagram showing Maternal Drug Intake distribution

Table 8: Admission

Admission	N=35	%
NICU Admission	6	17.1%
NO NICU Admission	29	82.9%

In the study 17.1% required NICU admission and 82.9% did not require NICU admission.

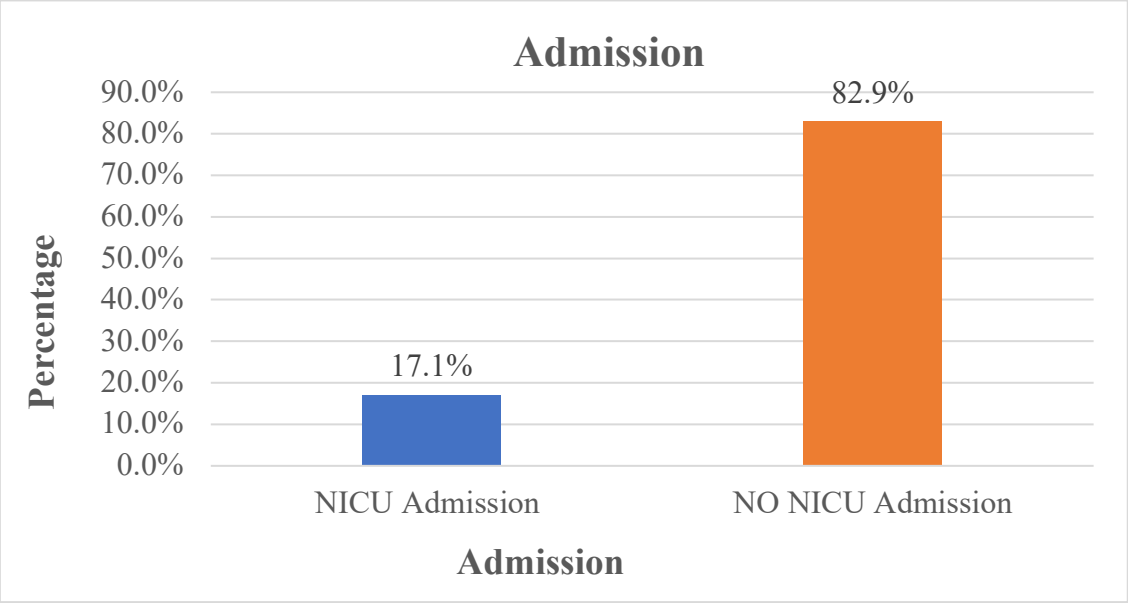

Figure 8: Bar diagram showing Admission

Table 9: Folic acid supplementation in mother

Folic Acid Supplementation	N=35	%
Taken	26	74.3%
Not taken	9	25.7%

In the study 74.3% had taken Folic Acid Supplementation and 25.7% did not take Folic Acid Supplementation.

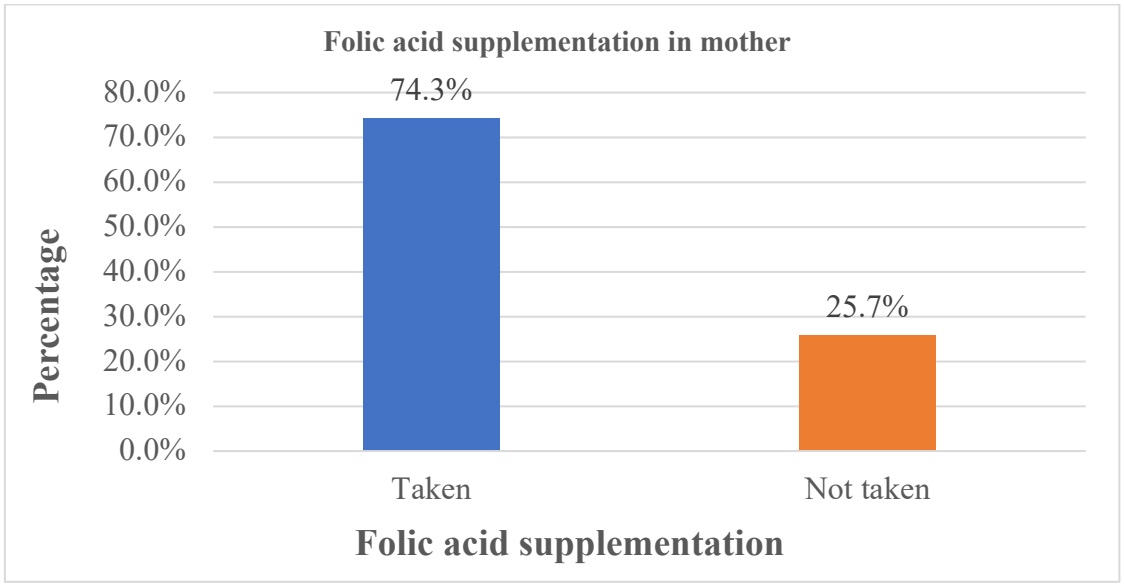

Figure 9: Bar diagram showing Folic acid supplementation in mother

Table 10: Age of the mother

Maternal Age	n = 35	%
20-25years	12	34.3%
26-30years	16	45.7%
31-35years	6	17.1%
36-40years	1	2.9%

In the study 34.2% of mothers were in the age group 20-25 years, 45.7% were in the age group 26-30 years, 17.1% were in the age group 31-35 years and 2.9% were in the age group 36-40 years. Mean Maternal age was 27.63 ± 3.889 years.

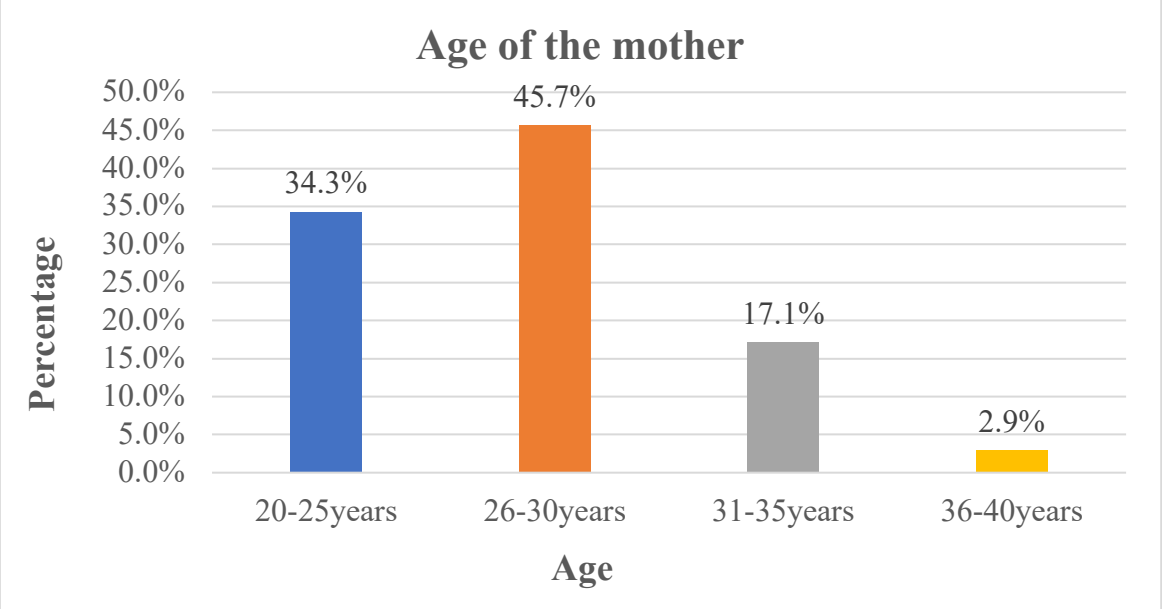

Figure 10: Bar diagram showing Age of the mother

Table 11: Parity of the mother

Parity	N=35	%
1	16	45.7%
2-3	17	48.6%
≥4	2	5.7%

In the study 45.7% of subjects were primiparous, 48.6% had parity of 2-3 and 5.7% had parity of ≥4.

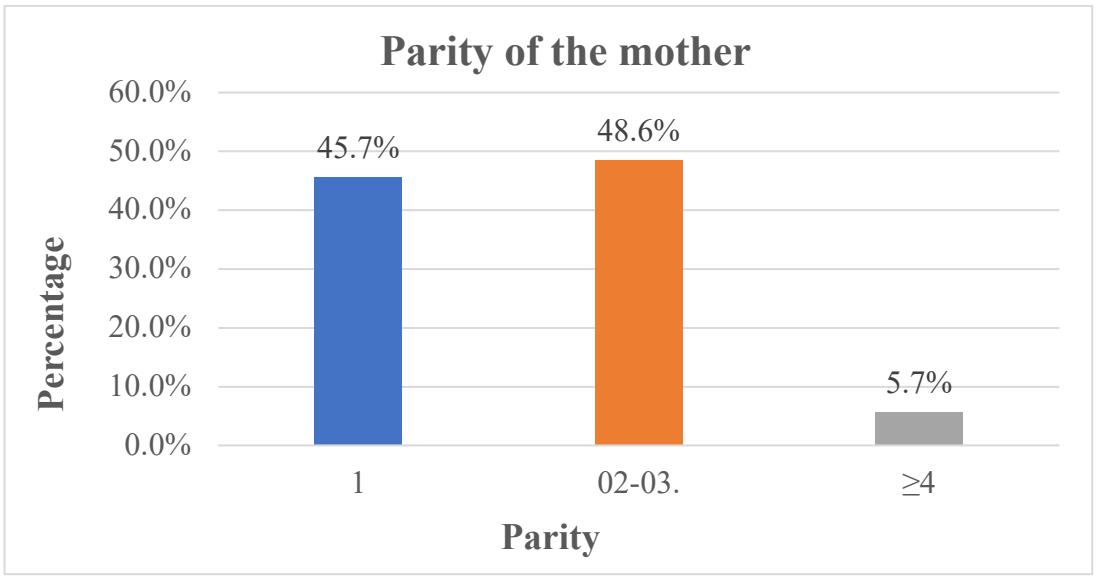

Figure 11: Bar diagram showing Parity of the mother

Table 12: Maternal Comorbidities

		Count	%
Maternal Comorbidities	Nil	29	82.9%
	PIH	3	8.6%
	GDM	3	8.6%

In the study 8.6% had PIH and GDM respectively.

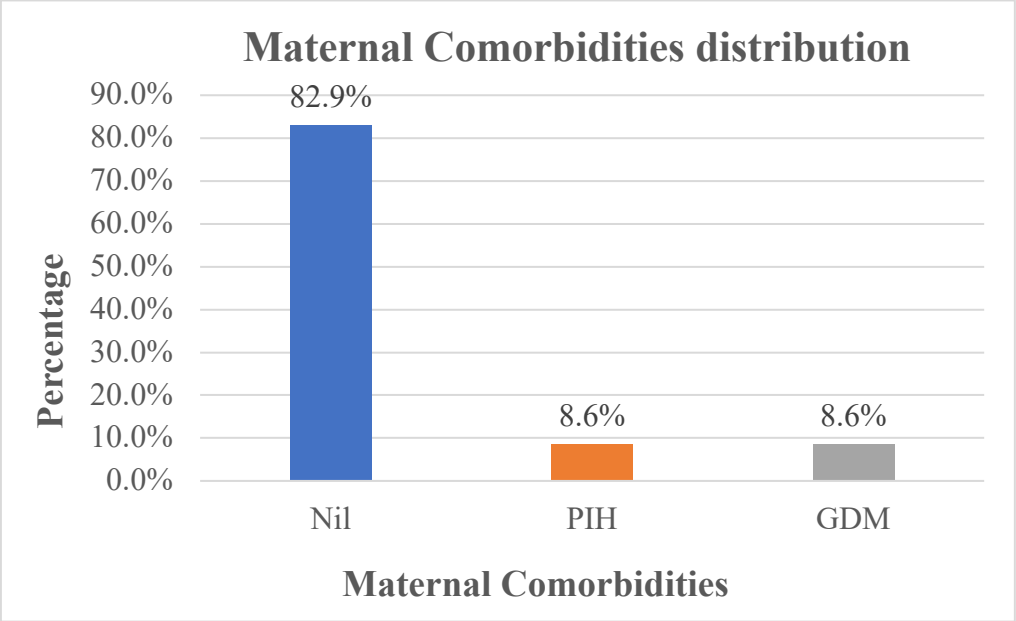

Figure 12: Bar diagram showing Maternal Comorbidities

Table 13: Mode of delivery distribution

Mode of delivery	N=35	%
Normal Vaginal	24	68.6%
Caesarean delivery	11	31.4%

In the study 68.6% of them were delivered by Normal Vaginal delivery and 31.4% were delivered by Caesarean delivery.

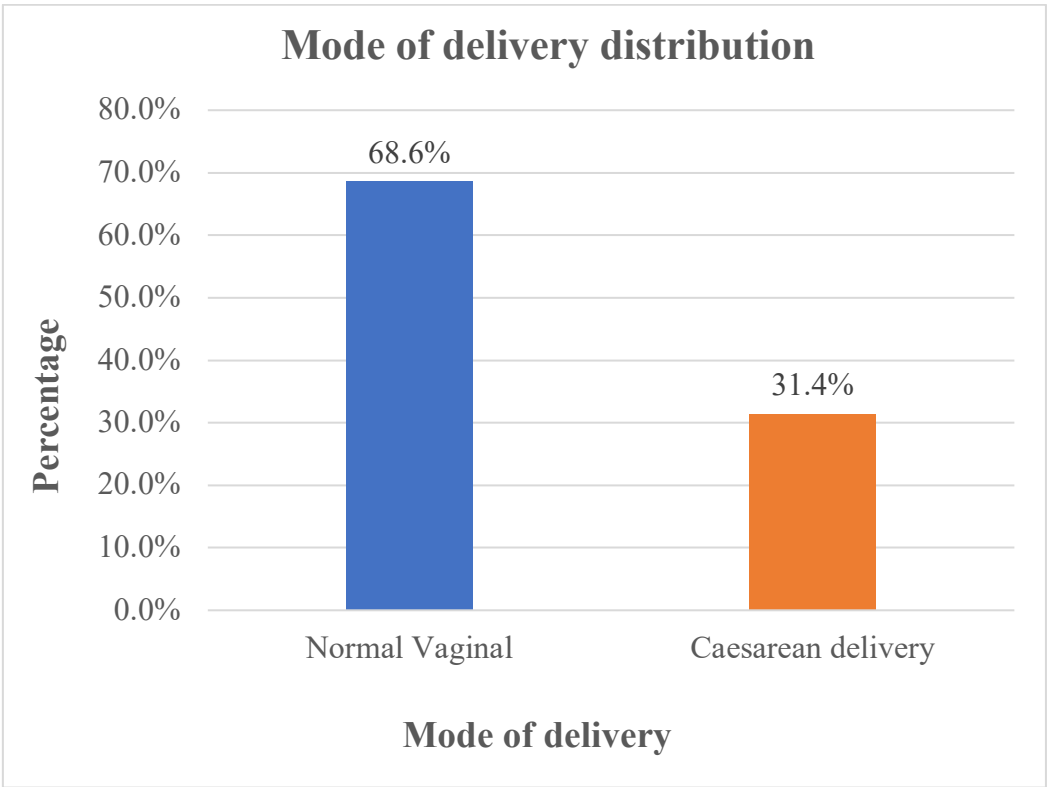

Figure 13: Bar diagram showing Mode of delivery distribution

Table 14: Phenotype distribution

			Count	%
Phenotype	Chromosomal Defects	Down Syndrome	6	17.2%
		Turners Syndrome	1	2.9%
	Genito Urinary	Anorectal Malformation	4	11.5%
		Ambiguous Genitilia	2	5.7%
		Cong Hydronephrosis	3	8.6%
		Hypospadias	3	8.6%
		Cong Urethral Stricture	1	2.9%
	GIT	Hirschsprung's Disease	2	5.7%
		Jejunal Atresia	1	2.9%
	Musculo Skeletal	Polydactyl	1	2.9%
		Syndactyl	1	2.9%
		Polysyndactyl	1	2.9%
	CVS	ASD	2	5.7%
		VSD	2	5.7%
	CNS	Occipital Encephalocoele	2	5.7%
	ENT	Microtia Right Ear	2	5.7%
	Skin	Preauricular Skin Tag	1	2.9%

In the study majority of them had phenotype of Down syndrome (17.2%), followed by Anorectal Malformation (11.5%) and Cong Hydronephrosis (8.6%) respectively and others as shown in above table.

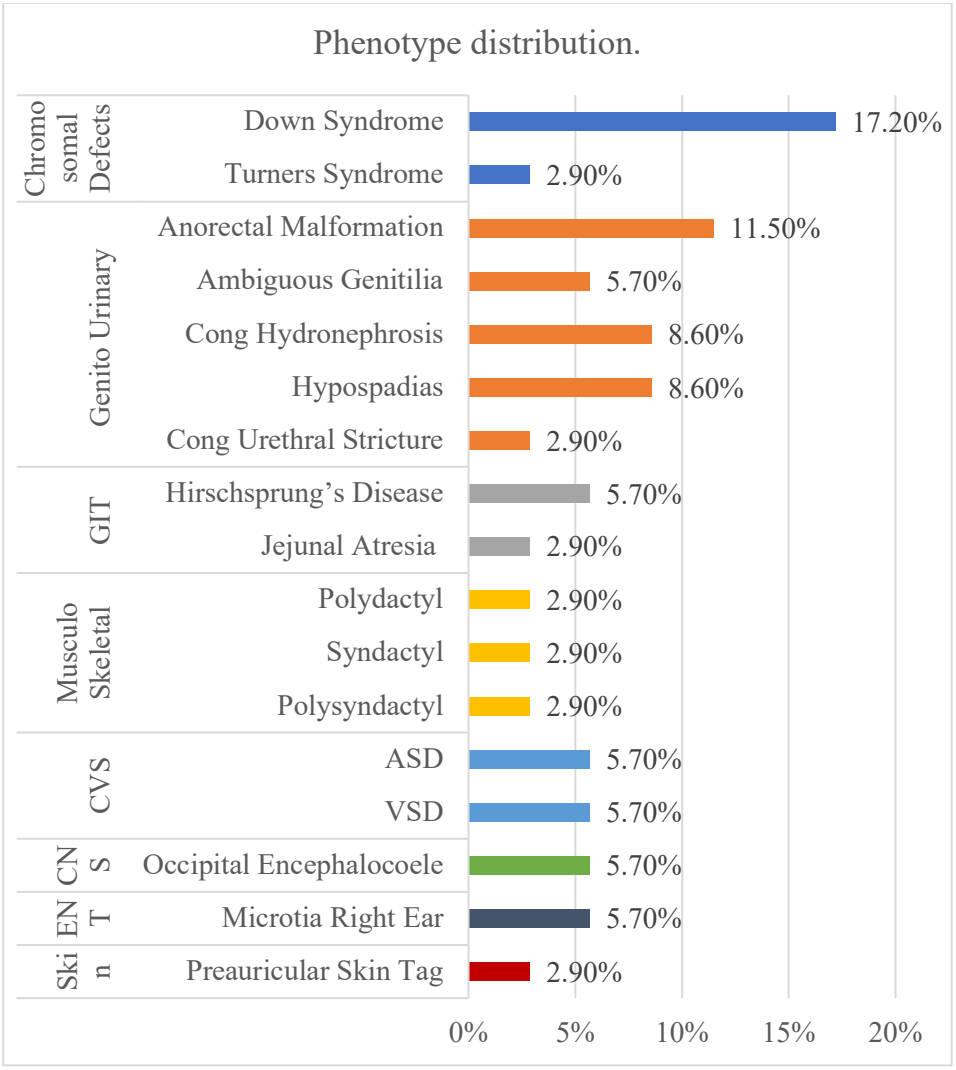

Figure 14: Bar diagram showing Phenotype distribution

Table 15: Karyotype distribution

		Count	%
Karyotype	46,XX	16	45.70%
	46,XY	7	20.00%
	46,XY,9qh+	5	14.30%
	47,XX,+21	3	8.60%
	47,XY,+21	2	5.70%
	46,XX,9qh+	1	2.90%
	46,XY,9qh+ 22pstk+	1	2.90%

In the study majority of the subjects had karyotype of 46,XX (45.7%), followed by 46,XY (20%), 46,XY,9qh+ (14.3%), 47,XX,+21 (8.6%), 47,XY,+21 (5.7%), 46,XX,9qh+ and 46,XY,9qh+ 22pstk+ (2.9%) respectively.

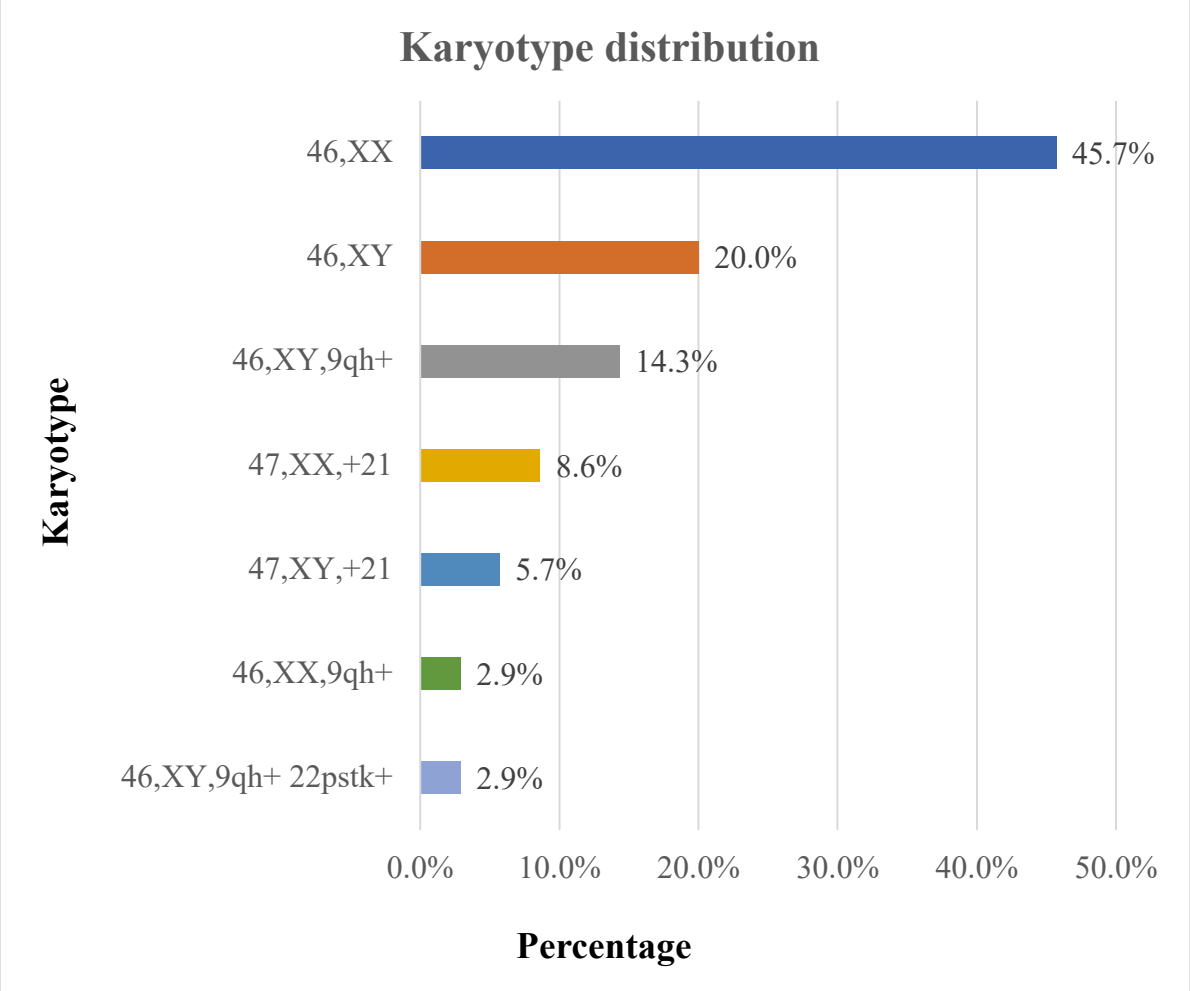

Figure 15: Bar diagram showing Karyotype distribution

Table 16: ICD Code

		Count	%
ICD Code	Q01.2	2	5.7%
	Q17	1	2.9%
	Q17.2	2	5.7%
	Q21.0	2	5.7%
	Q21.1	2	5.7%
	Q41.1	1	2.9%
	Q42.3	4	11.4%
	Q43.1	2	5.7%
	Q54.9	3	8.6%
	Q56	2	5.7%

	Q62.0	3	8.6%
	Q64.33	1	2.9%
	Q69	2	5.7%
	Q70.2	1	2.9%
	Q90	6	17.1%
	Q96	1	2.9%

In the study majority of subjects had ICD code of Q90 (17.1%), followed by Q42.3 in 11.4% and others as shown in above table.

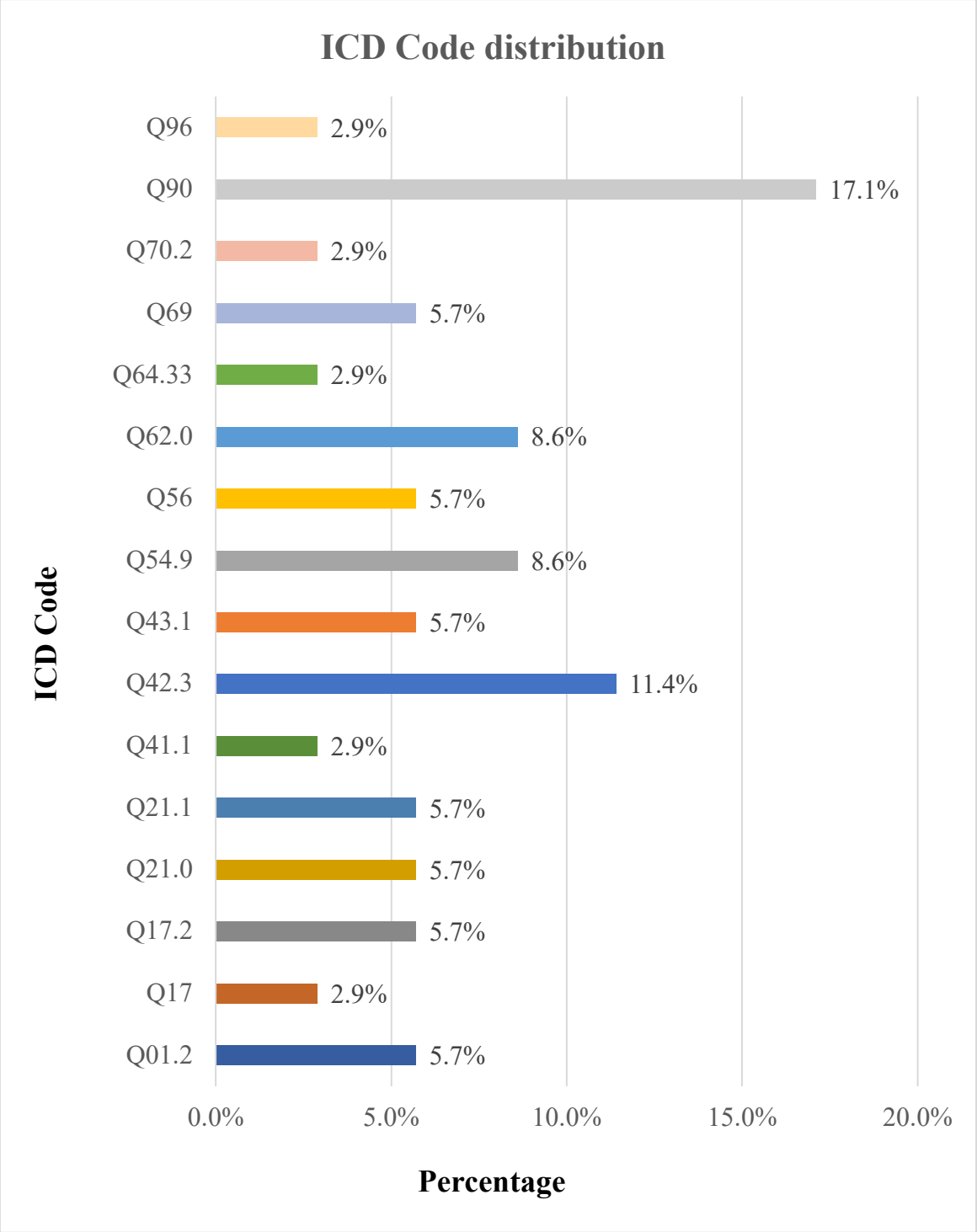

Figure 16: Bar diagram showing ICD Code

Table 17: Karyotyping distribution

		Count	%
Karyotyping	Normal	23	65.7%
	Abnormal	12	34.3%
	Total	35	100.0%

In the study 65.7% had normal Karyotyping and 34.3% had Abnormal Karyotyping.

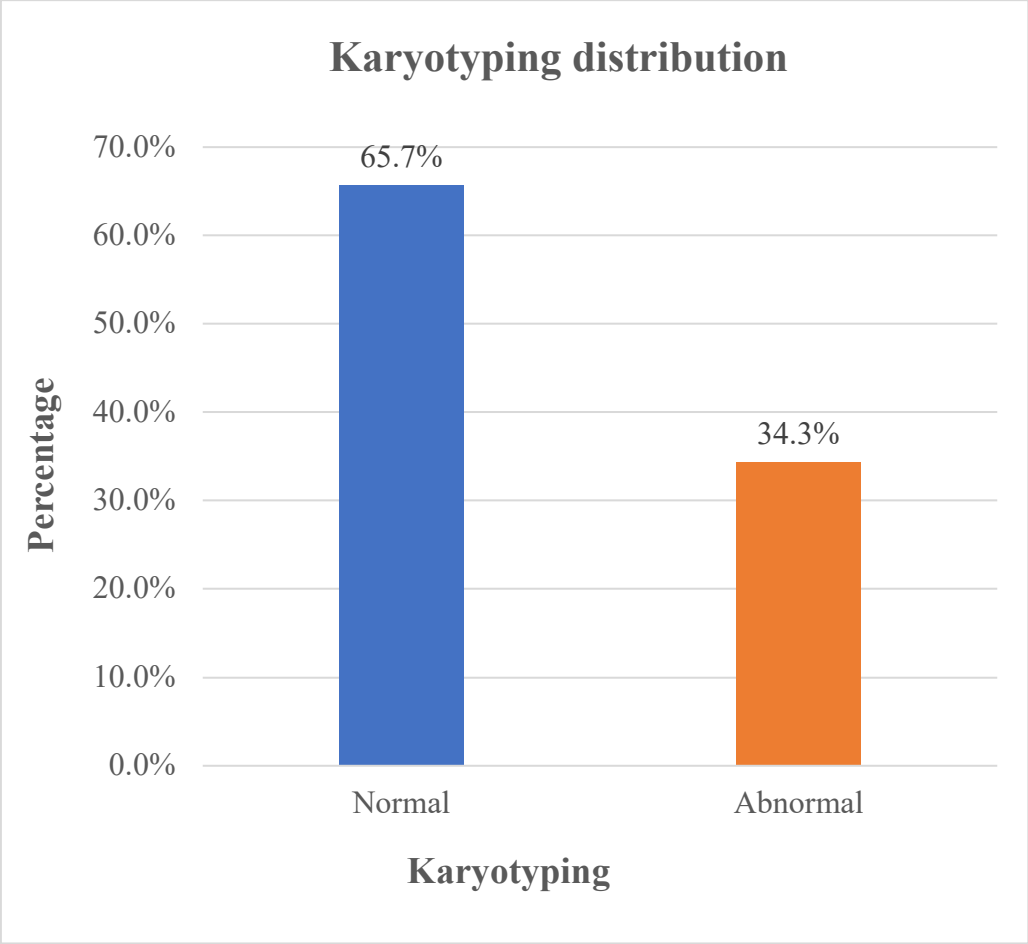

Figure 17: Bar diagram showing Karyotyping distribution

Table 18: Effects of associated fetal factors on percentage of congenital anomalies with Abnormal Karyotyping

		Karyotyping					P value
		Normal		Abnormal		Total	
		Count	Row N %	Count	Row N %	Count	
Gestation	Preterm	1	33.3%	2	66.7%	3	0.217
	Term	22	68.8%	10	31.2%	32	
Gender	Female	16	80.0%	4	20.0%	20	0.040*
	Male	7	46.7%	8	53.3%	15	
ICU Admission	No	20	69.0%	9	31.0%	29	0.373
	Yes	3	50.0%	3	50.0%	6	

Pearson Chi-Square Tests

In the study among preterm, 66.7% had abnormal karyotyping and 33.3% had normal karyotyping. Among term pregnancy, 31.1% had abnormal karyotyping and 68.8% had normal karyotyping. There was no significant association between Gestational period and karyotyping. Among Females, 20% had abnormal karyotyping and among males, 53.3% had abnormal karyotyping. There was significant association between Gender and karyotyping. Abnormal karyotyping was common among males with congenital anomalies.

Among subjects who required NICU admission, 50% had abnormal karyotyping and among subjects who did not require NICU admission, 31% had abnormal karyotyping. There was no significant association between Karyotyping and NICU admission.

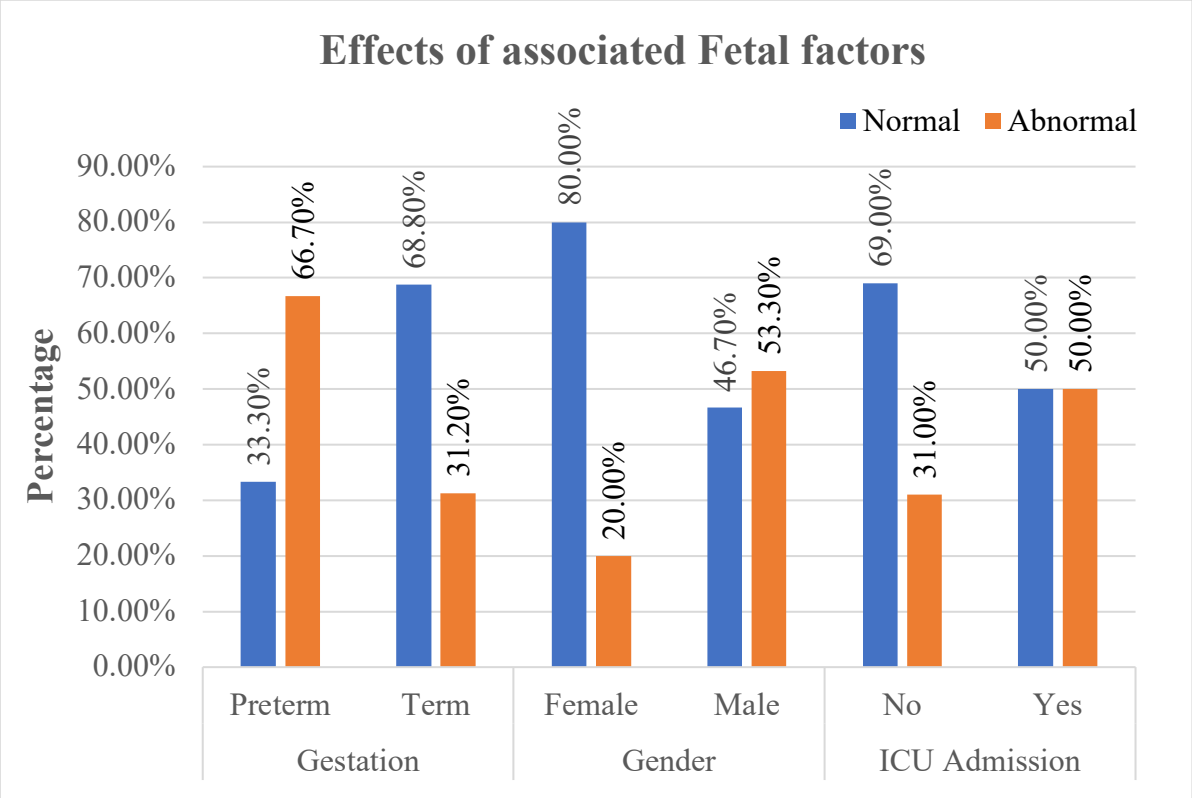

Figure 18: Bar diagram showing Effects of associated Fetal factors on percentage of congenital anomalies with Abnormal Karyotyping

Table 19: Effects of maternal factors on percentage of congenital anomalies with Abnormal Karyotyping

		Karyotyping					P value
		Normal		Abnormal		Total	
		Count	Row N %	Count	Row N %	Count	
Maternal Age	<35 years	23	67.6%	11	32.4%	34	0.160
	>35 years	0	0.0%	1	100.0%	1	
Parity	<4	22	66.7%	11	33.3%	33	0.630
	≥4	1	50.0%	1	50.0%	2	
Mode of Delivery	LSCS	7	63.6%	4	36.4%	11	0.421
	NVD	16	66.7%	8	33.3%	24	
Maternal Drug Intake	No	18	64.3%	10	35.7%	28	0.861
	Yes	5	71.4%	2	28.6%	7	
Consanguinity	Yes	6	25.3%	18	74.7%	24	0.04*
	No	9	81.0%	2	18.0%	11	
Oligo/Polyhydramnios	Polyhydramnios	1	100.0%	0	0.0%	1	0.464
H/O Congenital Anomaly	No	23	67.6%	11	32.4%	34	0.160
	Yes	0	0.0%	1	100.0%	1	

Pearson Chi-Square Tests

In the study among subjects aged >35 years, 100% had abnormal karyotyping, among subjects with parity <4, 33.3% had abnormal karyotyping and among subjects with parity ≥4, 50% had abnormal karyotyping. Among subjects who were delivered by LSCS, 36.4% had abnormal karyotyping, among subjects who were delivered by Normal vaginal delivery, 33.3% had abnormal karyotyping. Among mothers with history of Drug Intake, 28.6% had abnormal karyotyping among subjects without history of Drug Intake, 35.7% had abnormal karyotyping. Among subjects with Consanguinity, 74.7% had abnormal karyotyping. Among subjects with Non-consanguinity, 18% had abnormal

karyotyping. Among subjects with Polyhydramnios, 0% had abnormal karyotyping. Among subjects with H/O Congenital Anomaly, 100% of them had Abnormal karyotyping.

In the study Parental Consanguinity was significantly associated with abnormal karyotyping among subjects with congenital anomaly.

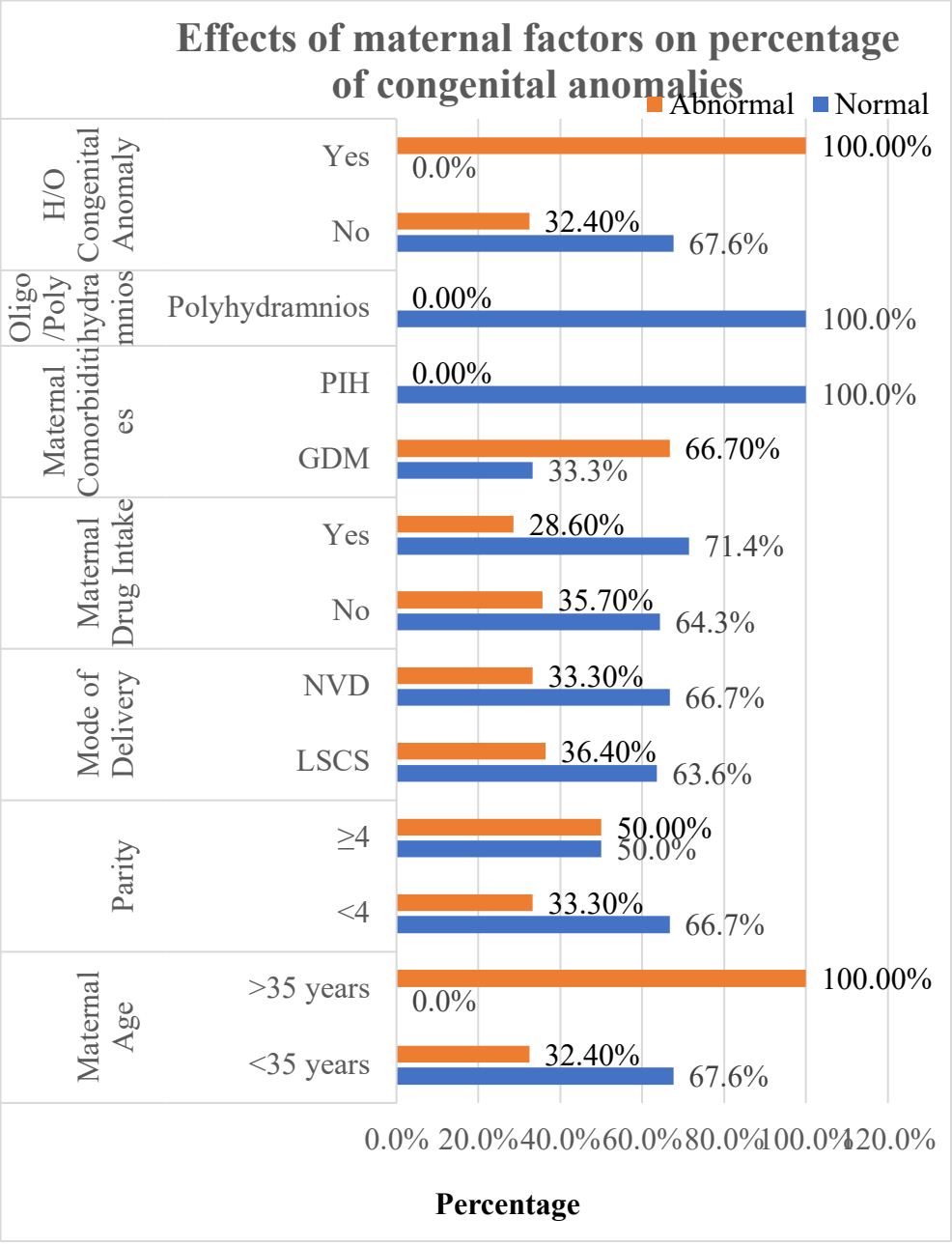

Figure 19: Bar diagram showing Effects of maternal factors on percentage of congenital anomalies with Abnormal Karyotyping

Discussion

The total of 35 patients with congenital anomaly who attended our medical college and gave informed consent were included in the study. Peripheral blood from these cases was collected and fifteen–twenty metaphase spread was examined under trinocular research microscope. Congenital anomalies were classified using ICD-10 classification. Type of abnormality and karyotype of the cases were the outcome variables.

In our study, 12 babies had abnormal karyotyping (34.3%) and 23 babies had normal karyotyping (65.7%). Abnormal karyotyping being 46,XY,9qh+ (14.3%), 47,XX,+21 (8.6%), 47,XY,+21 (5.7%), 46,XX,9qh+ and 46,XY,9qh+ 22pstk+ (2.9%) .

In our study, chromosomal abnormality mainly Downs Syndrome was seen with karyotype results being 47,XX,+21(8.6%) and 47,XY,+21(5.7%) which was comparable with the study done by a retrospective analytical study conducted by Kava et al, [28] included a sample of 524 patients in which males contributed majorly with 57.8% and females of 42.1% which is in great agreement with our study proportions of 57.1% males and 42.9% females.

With regard to pattern of congenital anomalies in the study, the most common system involved was genitourinary (37.14%), followed by musculoskeletal system (17%), CVS (11.4%), gastrointestinal (GIT) (8.6%), central nervous system(CNS) (5.7%), skin (2.8%) etc. whereas Suguna Bai et al. [29] reported GI malformations as the most common one. In a study done by Mohamed A. El Koumi et al, [30] the most common system involved was the musculoskeletal system, followed by the CNS.

Suguna Bai et al. [29] reported a higher incidence of malformation in the babies born to mothers aged over 35(84) years, whereas Dutta et al. [31] documented statistically insignificant association of increased maternal age and congenital anomalies. In our study, the relationship between maternal age and babies born with congenital malformations revealed that a majority of malformed babies were born of mothers aged 26-30 years; though, it was statistically insignificant. On the other hand, Anjum et al. [32] reported that the majority of neonates with Congenital Anomalies are born to mothers aged 25-38 years.

Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations. In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages as seen in study done by Madi SA et al. [33] Our result was consistent with the result seen in the study done by Mohamed A. El Koumi et al [30] which stated Congenital Anomalies prevailed in babies of consanguineous marriage compared to non-consanguineous marriage.

In our study, higher incidence of malformations was seen among the multiparous women. Our result is consistent with the finding seen in the study done by Mohanty C et al. [34] which indicates a positive correlation between the birth order and the incidence of congenital anomalies.

In our study the incidence of congenital anomalies was significantly higher in term babies as compared with the preterm babies which was inconsistent with the study done by Mathur B C et al. [35] which showed the incidence of congenital anomalies was significantly higher in preterm babies as compared with the full-term babies. Also in a study done by Mohamed A. El Koumi et al, [30] Congenital Anomalies were observed significantly more in preterm babies than full term. Jones et al, [36] added that the risk factors associated with prematurity has proven increased frequency of Congenital Anomalies.

In our study, the mode of delivery did not show any significant association with congenital anomalies, whereas in a study done by Shatanik Sarkar et al. [10] showed significant association with congenital anomalies with caesarean section being more commonly associated than normal delivery.

In a study done by Shatanik Sarkar et al., [10] male babies had significant association with congenital anomalies than females, which was also seen in our study where male babies had significant association with congenital malformation than females. But in a study done by Mohamed A. El Koumi et al [30] there was no significant difference in the frequency of Congenital Anomalies in male rather than female babies. On the other hand, Gupta et al. [37] reported that the incidence of congenital musculoskeletal malformations was apparently found to be higher in female babies than in males; however, the difference was not statistically significant.

In a study done by Mohamed A. El Koumi et al, [30] there were significantly more cases with a history of an anomaly in another child or in the family among mothers of neonates with Congenital Anomaly. In the present study 2.9% had family history of congenital anomaly.

In a study done by Hernandez-Diaz et al, [38] showed that periconceptional intake of medications acting as folic acid antagonists, including anti-epileptic agents and dihydrofolate reductase inhibitors, doubled the risk of cardiovascular defects. In the present study only 7(20%) patients had history of maternal drug intake among which 3 cases had reported with congenital anomaly. Tang et al, [39] demonstrated that impaired folic acid transport results in extensive apoptotic cell death in the developing heart; apoptotic cells were shown to be concentrated in the truncus arteriosus and interventricular septum and were thus anatomically restricted to the two regions in which most congenital heart defects are found.

In a study done by Amsalu Taye Wondemagegn et al, [40] revealed that through supplementation of folic acid immediately before and in the early period of pregnancy, the risk of CHDs of various type could be reduced by 21%.

In our study 74.3% of women [26] had taken folic acid supplementation and 25.7% [9] did not take folic acid supplementation.

Limitations:

Study was conducted in a single hospital-based sample with less sample size.

Conclusion:

- Anomalies were most likely to be in the genitourinary system.
- Maternal history of parental consanguinity was significantly associated with an increased risk of congenital anomalies. In the study 68.6% had non-consanguineous marriage and 31.4% had consanguineous marriage.
- There was no significant association between Gestational period and karyotyping
- There was significant association between Gender and karyotyping. Abnormal karyotyping was common among males with congenital anomalies.
- Supplementation with folic acid in early period of pregnancy reduces the risk of CHD's and various CNS malformations. In our study majority of women had taken folic acid supplementation.
- Malformation scan can detect lethal congenital anomalies. Antenatal testing like amniotic fluid testing can be used to detect certain lethal congenital anomalies, hence maternal education and family planning play a very important role in prevention of congenital anomalies. Consanguinity is associated with increased incidence of anomalies so it should be discouraged.
- Adequate antenatal care was significantly associated with a decreased risk.
- No statistically significant associations were found for other neonatal factors.
- Genetic counselling plays a vital role for the high-risk parents. It provides information regarding various procedure and diagnostic technique, the risk of consequences of some of the procedure, as well as about options available. Genetic counselling at different time period helps in reduction of congenital anomalies, morbidity and mortality resulting from these anomalies
- Public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended.
- Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

Reference

1. BK Malla. One year review study of congenital anatomical malformation at birth in Maternity Hospital (Prasutigriha), Thapathali, Kathmandu - PubMed [Internet]. [cited 2024 Jun 9]. Available from: https://pubmed.ncbi.nlm.nih.gov/18604096/
2. Carter CO. Congenital Defects: The Genetics of Congenital Malformations. J R Soc Med. 1968;61(10):991–5.
3. Martin RM, Fanaroff AA, Walsh MC. Fanaroff and Martin's neonatal-perinatal medicine diseases of the fetus and infant, 8th edn, Vols I and II. Arch Dis Child Fetal Neonatal Ed [Internet]. 2006 [cited 2024 Jun 9];91(6):F468. Available from: /pmc/articles/PMC2672777/
4. Ali A, Zahad S, Masoumeh A, Azar A, Khominee Hospital E, Ahmadzadeh Ali C. CONGENITAL MALFORMATIONS AMONG LIVE BIRTHS AT ARVAND HOSPITAL, AHWAZ, IRAN-A PROSPECTIVE STUDY. Pak J Med Sci [Internet]. 2008 [cited 2024 Jun 9];24(1):33–7. Available from: www.pjms.com.pk33
5. Grover N. Congenital malformations in Shimla. Indian J Pediatr [Internet]. 2000 [cited 2024 Jun 9];67(4):249–51. Available from: https://pubmed.ncbi.nlm.nih.gov/10878862/
6. Patel ZM, Adhia RA. Birth defects surveillance study. Indian J Pediatr [Internet]. 2005 [cited 2024 Jun 9];72(6):489–91. Available from: https://pubmed.ncbi.nlm.nih.gov/15985737/
7. WHO. Congenital disorders [Internet]. [cited 2024 Jun 9]. Available from: https://www.who.int/health-topics/congenital-anomalies#tab=tab_1
8. Ravi Kumar M, Vishnu Bhat B, Oumachigui A. Perinatal mortality trends in a referral hospital. Indian J Pediatr [Internet]. 1996 [cited 2024 Jun 9];63(3):357–61. Available from: https://pubmed.ncbi.nlm.nih.gov/10830011/
9. SS Agarwal. Prevalence & spectrum of congenital malformations in a prospective study at a teaching hospital - PubMed [Internet]. 1991 [cited 2024 Jun 9]. Available from:https://pubmed.ncbi.nlm.nih.gov/1774092/
10. Shatanik Sarkar. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India - PubMed [Internet]. [cited 2024 Jun 9]. Available from: https://pubmed.ncbi.nlm.nih.gov/24251257/
11. Sadler TW Langman. Langman's Medical Embryology 14th Edition PDF | Vet eBooks [Internet]. [cited 2024 Jun 9]. Available from: https://www.vet-ebooks.com/langmans-medical-embryology-14th-edition-pdf/
12. Thottumkal. Thottumkal VA, Deepak M. INTERNATIONAL JOURNAL OF... - Google Scholar [Internet]. [cited 2024 Jun 9]. Available from: https://scholar.google.com/scholar?hl=en&as_sdt=0%2C5&q=Thottumkal+VA%2C+Deepak+M.+INTERNATIONAL+J

OURNAL+OF+PHARMACEUTICAL%2C+CHEMICAL+AND+BIOLOGICAL+SCIENCES+CONGENITAL+ANOMALIES&btnG=
13. Boris. Nelson Textbook of Pediatrics, 2-Volume Set - 9780323529501 | Elsevier Health [Internet]. [cited 2024 Jun 9]. Available from: https://www.mea.elsevierhealth.com/nelson-textbook-of-pediatrics-2-volume-set-9780323529501.html
14. Ameen SK, Alalaf SK, Shabila NP. Pattern of congenital anomalies at birth and their correlations with maternal characteristics in the maternity teaching hospital, Erbil city, Iraq. BMC Pregnancy Childbirth [Internet]. 2018 Dec 18 [cited 2024 Jun 9];18(1):1–8. Available from:https://bmcpregnancychildbirth.biomedcentral.com/articles/10.1186/s12884-018-2141-2
15. Al-Gazali LI, Dawodu AH, Sabarinathan K, Varghese M. The profile of major congenital abnormalities in the United Arab Emirates (UAE) population. J Med Genet [Internet]. 1995 [cited 2024 Jun 7];32(1):7–13. Available from:https://pubmed.ncbi.nlm.nih.gov/7897633/
16. Sawardekar KP. Profile of major congenital malformations at Nizwa Hospital, Oman: 10-year review. J Paediatr Child Health [Internet]. 2005 Jul [cited 2024 Jun 9];41(7):323–30. Available from: https://pubmed.ncbi.nlm.nih.gov/16014135/
17. Walden R V., Taylor SC, Hansen NI, Poole WK, Stoll BJ, Abuelo D, et al. Major congenital anomalies place extremely low birth weight infants at higher risk for poor growth and developmental outcomes. Pediatrics [Internet]. 2007 Dec [cited 2024 Jun 9];120(6). Available from: https://pubmed.ncbi.nlm.nih.gov/17984212/
18. Anyanwu. Birth Prevalence of Overt Congenital Anomalies in Kano Metropolis: Overt Congenital Anomalies in the Kano [Internet]. [cited 2024 Jun 9]. Available from: https://www.researchgate.net/publication/283185968_Birth_Prevalence_of_Overt_Congenital_Anomalies_in_Kano_Metropolis_Overt_Congenital_Anomalies_in_the_Kano
19. Habib. The incidence of congenital malformation and associated... - Google Scholar [Internet]. [cited 2024 Jun 9]. Available from: https://scholar.google.com/scholar?hl=en&as_sdt=0%2C5&q=The+incidence+of+congenital+malformation+and+associated+risk+factors+in+Basrah+city&btnG=
20. Christianson. March of Dimes: global report on birth defects, the hidden toll of dying and disabled children. | Request PDF [Internet]. [cited 2024 Jun 9]. Available from: https://www.researchgate.net/publication/32890332_March_of_Dimes_global_report_on_birth_defects_the_hidden_toll_of_dying_and_disabled_children
21. Singh A. Pattern of Congenital Anomalies in Newborn [Internet]. Available from: www.jkscience.org
22. Sharma R. Birth defects in India: Hidden truth, need for urgent attention. Indian J Hum Genet [Internet]. 2013 [cited 2024 Jun 9];19(2):125. Available from: https://pubmed.ncbi.nlm.nih.gov/24019610/
23. Neelambari YC, Das P, Sadagopan S, Uma AN. Prevalence, pattern and outcome of congenital malformations in a tertiary care centre in South India. Int J Contemp Pediatrics [Internet]. 2018 Apr 20 [cited 2024 Jun 9];5(3):1044–8. Available from: https://www.ijpediatrics.com/index.php/ijcp/article/view/1489
24. Dastgiri S, Stone DH, Le-Ha C, Gilmour WH. Prevalence and secular trend of congenital anomalies in Glasgow, UK. Arch Dis Child [Internet]. 2002 [cited 2024 Jun 9];86(4):257–63. Available from: https://pubmed.ncbi.nlm.nih.gov/11919098/
25. Park K. Park's Text Book of Preventive and Social Medicine (PSM) – 26th Edition (2021). 2021; 2021.
26. Merchant. Merchant SM. Indian Council of Medical Research.... - Google Scholar [Internet]. [cited 2024 Jun 9]. Available from: https://scholar.google.com/scholar?hl=en&as_sdt=0%2C5&q=Merchant+SM.+Indian+Council+of+Medical+Research.+Genetic+Research+Centre%2C+Bombay%2C+Annual+Report+1989&btnG=
27. Singh M. Hospital-based data on perinatal and neonatal mortality in India - PubMed [Internet]. [cited 2024 Jun 9]. Available from: https://pubmed.ncbi.nlm.nih.gov/3643164/
28. Kava MP, Tullu MS, Muranjan MN, Girisha KM. Down syndrome: clinical profile from India. Arch Med Res [Internet]. 2004 [cited 2024 Jun 7];35(1):31–5. Available from: https://pubmed.ncbi.nlm.nih.gov/15036797/
29. N S Suguna Bai. An etiological study of congenital malformation in the newborn - PubMed [Internet]. [cited 2024 Jun 7]. Available from: https://pubmed.ncbi.nlm.nih.gov/7160886/
30. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: A hospital-based study. Pediatr Rep. 2013;5(1):20–3.
31. Vikram Dutta. Indian Pediatrics - Editorial [Internet]. [cited 2024 Jun 7]. Available from: https://www.indianpediatrics.net/sept2000/sept-998-1001.htm
32. Anjum R, Saher F, Nargis S. Frequency of congenital anomalies and associated maternal risk factors in the lower socio-economic group. 2006;169–73.

33. S A Madi. Profile of major congenital malformations in neonates in Al-Jahra region of Kuwait - PubMed [Internet]. [cited 2024 Jun 7]. Available from: https://pubmed.ncbi.nlm.nih.gov/16700386/
34. Mohanty. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G.... - Google Scholar [Internet]. [cited 2024 Jun 9]. Available from: https://scholar.google.com/scholar?hl=en&as_sdt=0%2C5&q=Mohanty+C%2C+Mishra+OP%2C+Das+BK%2C+Bhatia+BD%2C+Singh+G.+Congenital+malformations+in+newborns%3A+A+study+of+10%2C874+consecutive+births.+J+Anat+Soc+India+&btnG=
35. B C Mathur. Congenital malformations in the newborn - PubMed [Internet]. [cited 2024 Jun 7]. Available from: https://pubmed.ncbi.nlm.nih.gov/1171821/
36. Jones. Smith's Recognizable Patterns of Human Malformation - E-Book - Kenneth Lyons Jones, Marilyn Crandall Jones, Miguel del Campo - Google Books [Internet]. [cited 2024 Jun 9]. Available from: https://books.google.co.in/books?hl=en&lr=&id=5wEhEAAAQBAJ&oi=fnd&pg=PP1&dq=Jones+KL.+Smith%27s+recognizable+patterns+of+human+malformation.+Philadelphia:+WB&ots=XPCX36dY5P&sig=9_Vqa0BygxrRXjKhBcDwIFAeexc#v=onepage&q&f=false
37. Gupta RK, Gupta CR, Singh D. Incidence of Congenital Malformations of the MusculoSkeletal System in New Live Borns in Jammu. 2003 [cited 2024 Jun 9]; Available from:http://imsear.searo.who.int/handle/123456789/171055
38. Hernández-Díaz S, Werler MM, Walker AM, Mitchell AA. Folic acid antagonists during pregnancy and the risk of birth defects. N Engl J Med [Internet]. 2000 Nov 30 [cited 2024 Jun 7];343(22):1608–14. Available from: https://pubmed.ncbi.nlm.nih.gov/11096168/
39. Tang LS, Wlodarczyk BJ, Santillano DR, Miranda RC, Finnell RH. Developmental consequences of abnormal folate transport during murine heart morphogenesis. Birth Defects Res A Clin Mol Teratol [Internet]. 2004 Jul [cited 2024 Jun 7];70(7):449–58. Available from: https://pubmed.ncbi.nlm.nih.gov/15259034/
40. 40. Wondemagegn AT, Afework M. The association between folic acid supplementation and congenital heart defects: Systematic review and meta-analysis. SAGE Open Med [Internet]. 2022 Mar 1 [cited 2024 Jun 9];10. Available from: https://journals.sagepub.com/doi/full/10.1177/20503121221081069