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Prospective Observational Study on Prevalence and Outcome of Congenital Abnormalities in Fetus in Tertiary Health Care

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Abstract: ***Background: Congenital abnormalities are the abnormality of structure, function, or body metabolism present at birth and result in physical/mental disability or is fatal. Congenital anomalies are important cause of perinatal, early neonatal and infant morbidity and mortality. most of birth defects are preventable. The frequency of the anomalies can be reduced by either removing the risk factor or reinforcement of protective factors. Method: Prospective descriptive study; Sample size: (N=53); Overall 39 births (live +stillbirth) & 132 MTPs at tertiary health care to assess the prevalence and outcome of congenital abnormalities in fetus and to assess the most common congenital abnormalities in the tertiary health care. Result: Incidence of anomalous fetuses (live + stillbirth + MTPS) is 1.3; majority patients were 26-30yr (mean age 27.13) and incidence is more among multiparous than nulliparous and distributed more amongst the class 3 of modified kuppuswamy scale: frequent medical comorbidities were DM, HTN +DM, HTN, Asthma. Distributed more among non consanguineous marriages and nil history of prior anomalous birth and family history of congenital abnormal fetus. About 22.6% of the mother has inadequate intake of folic acid supplementation and 13% associated with CNS anomalies. The remaining mother had anomalous fetuses inspite of folic acid supplements reason being inadequate periconceptional FA supplementation. Major anomalies=68%, male prevalence=55% out of which 72.4% had major abnormalities. Total 61 anomalies were identified. CNS Anomalies=46%; cardiac malformation=23%; genitourinary+10% etc. Pregnancy outcome were Live birth=60%; MTP=32%. APGAR Score of >7 had good prognosis and were observed in NICU and NND=71%; stillbirth=29%. Conclusion: congenital abnormalities leading to developmenmtal abnormalities if diagnosed at an early gestational age can be terminated. Awareness of antenatal, postnatal care has decreased the perinatal mortality; however perinatal death due to congenital malformations still remains a major group, can be prevented by periconceptional folic acid supplementation. Congenital malformations due to preventable causes like alcohol; infections; FA supplements can be decreased by spreading awareness or educating via periconceptional counselling.***

Keywords: Congenital Anomalies, Stillbirth, MTPs, FA Supplementation, Periconceptional Counselling.

1. Introduction

Congenital abnormality is the abnormality of structure, function or body metabolism present at birth and results in physical or mental disability or is fatal. Congenital anomalies are important cause of perinatal, early neonatal and infant morbidity and mortality. One of the leading causes of infant morbidity and mortality in developed countries is congenital malformations. The increase in the incidence of congenital abnormalities are underestimated as the deficiency in diagnostic capabilities and lack of reliability. ICD-10 has classified the congenital abnormalities into Major and Minor defects. Major birth defects are grouped into External and Internal defects. Birth defects registry of India and foetal care research foundation enrolled neural tube defects as the most common anomaly.

Congenital malformations are confined to structural defects at birth. About 270000 newborn die every year within 28 days of birth worldwide due to congenital anomalies[1].

There have been multiple causes and risk factors for these anomalies being genetic, socioeconomic, demographic, environmental, infections, maternal nutritional status, consanguinity etc.

The prevalence of congenital abnormalities in India is 6-7% in 2006 March of Dimes report. In Europe the cause for perinatal mortality was 1.27 / 1000 births, average stillbirth rate was 2.68% as per EUROCAT[2]. In USA congenital abnormalities accounted 20% infant mortality and 2.3% of premature deaths and disability[3]. The prevalence of congenital anomalies varies across different parts of India like 2.3% in Pune, 1.9% in Tamilnadu, 2.38% in Ahmedabad[4], 1.2% in Mangalore[5] etc.

Most of the birth defects are preventable. The frequency of the anomalies can be reduced by either removing the risk factors or reinforcement of protective factors. Up to 70% of birth defects can be reduced by improving Diagnosis, care and prevention[6].

2. Aims and Objectives

1) To assess the prevalence of congenital abnormalities in fetus in tertiary healthcare centre.
2) To assess the most common congenital abnormality.

3) To assess the outcomes of congenital abnormalities of fetus in tertiary healthcare centre.

3. Materials and Methods

Study Design: a prospective descriptive study conducted at Tertiary referral centre.

Study Population: 3900 births (live births & stillbirths) and 132 MTPs

Study Duration: from February 2020 to August 2021

Study Setting: Obstetrics and Gynecology department OF LTMMC

Inclusion Criteria -
- All Pregnant women with USG scan suggestive of anomalous fetus
- Those consenting for the study

Exclusion Criteria -
- Not consenting for study

Data Collection:
All the patients with documented USG suggestive of anomalous fetus attending the Outpatient department and labor room of obstetrics and gynaecology department at Tertiary Referral Centre included in the study.

Written and informed consent taken.

Demographic details like maternal age, consanguinity, education, socioeconomic status, maternal infection, birth order, maternal nutrition, family history, medical history, obstetric history, exposure to drugs, personal history have been collected at their first visit and documented in the proforma.

Routine antenatal investigations were performed for all.

Specific investigations carried out as per the systemic diseases and obstetric needs / complications on case to case basis.

Detailed examination of abortus / fetus with congenital anomalies will be done.

4. Results

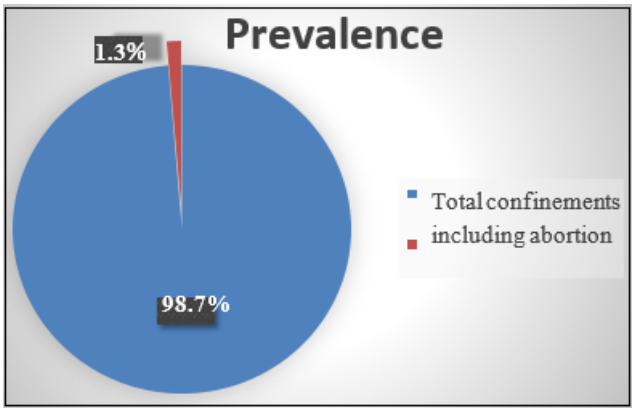

Figure 1: Pie Chart of Prevalence of Congenital anomalies

Outcome noted after collecting all the details and analyzed by SPSS-28 Software

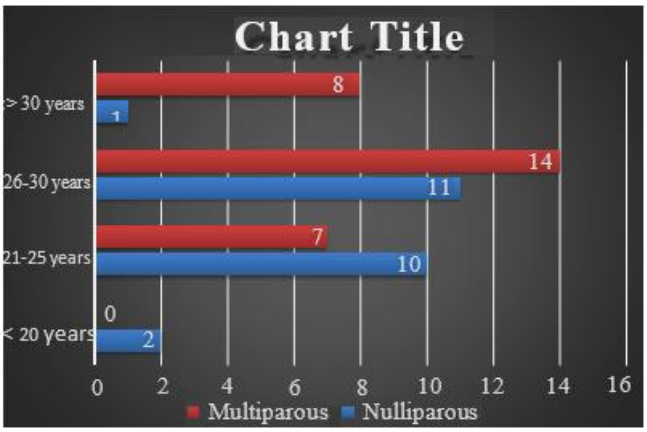

Figure 2: Graph representing Parity among study subjects

Out of 4032 patients delivered or aborted with us, 53 patients were found to have congenital anomalies; this accounted for a prevalence of 1.3%.

Our study has more number of congenital anomalies in the mothers of age group between 26-30 years among multiparous women.

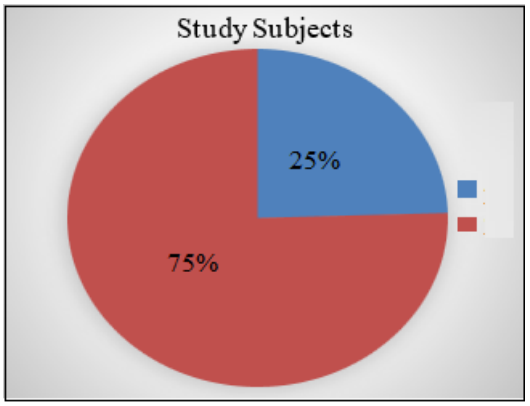

Figure 3: Graph representing Distribution according to socioeconomic

Status
75% of the families belonged to Class-3 of the Modified Kuppuswamy Scale in this study with the rest belonging to Class-2.

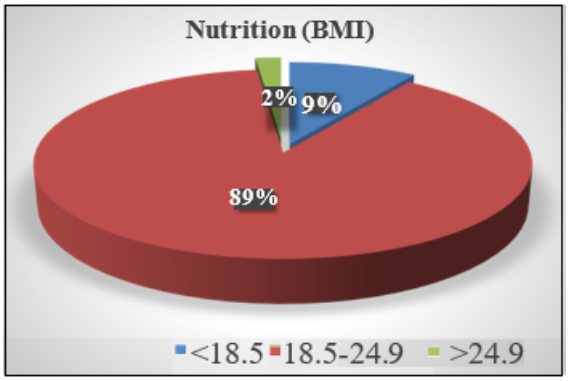

Figure 4: Graph representing Nutrition of the Mothers

88.7% of the mothers in our study were found to be in the BMI range of (18.5- 24.9 kg/ sq.m) followed by the mothers <18.5kg/sq.m (9.4%).

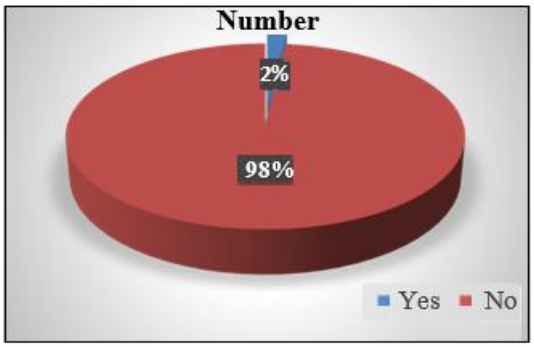

Figure 5: Graph representing Consanguineous Marriage

Our study had only 1 mother with a history of Consanguineous marriage.

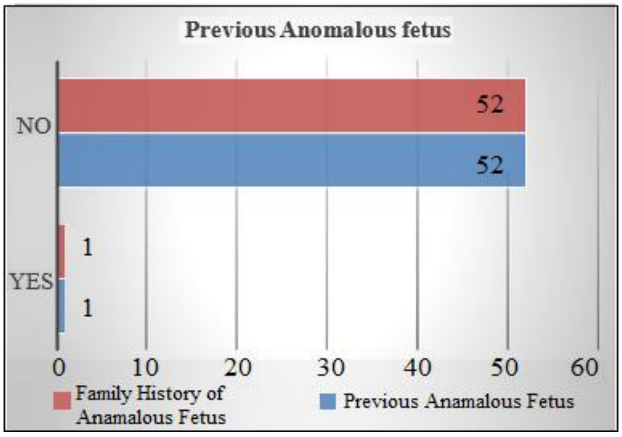

Figure 6: Graph representing Previous / Family Anomalous Fetus

One mother in our study have shown to have previous anomalous birth (Anencephaly); One mother had a Family history of anomalous births in 1st degree relative.

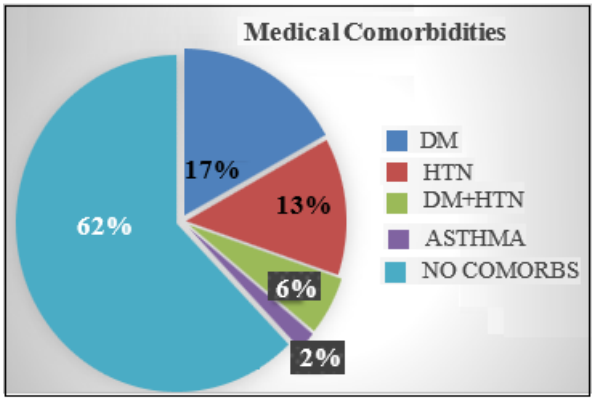

Figure 7: Graph representing medical illness to congenital anomalies

38% of the mothers had medical illnesses with 22.5% having Diabetes (isolated + combined) with commonly associated with CVS and CNS anomalies. 19% of the mothers had Hypertension (isolated + combined).

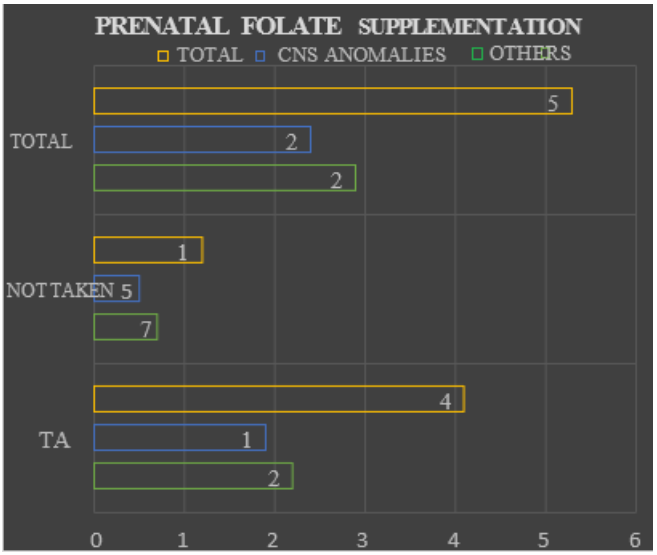

Figure 8: Graph representing Prenatal Folic acid supplementation

About 22.6% of the mothers had inadequate intake of Folic acid supplements with 13% associated with CNS anomalies. The remaining mothers had anomalous fetuses inspite of taking folic acid supplements, the reason being inadequate periconceptional folic acid supplementation.

The Major anomalies constitute 68% of the total anomalies. Males constitute 55% of the total anomalous fetuses out of which 72.4% had major anomalies, whereas 62.5% of the female fetuses had major congenital anomalies. Most of the mothers were in the age group of 26-30 years (47%) and 21-25 years (32.1%). The Mean age (SD) was found to be 27.13 (3.85) years.

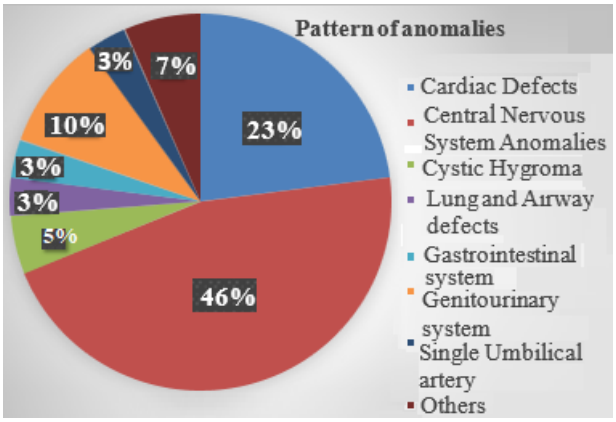

Figure 9: Graph representing the Pattern of congenital anomalies

61 Congenital anomalies identified in 53 pregnancies.

4 fetuses of the 53 had congenital anomalies in association with 45 fetuses in Isolation

There was a higher proportion of Major anomalies 39 (64%) compared to Minor anomalies 22 (36%)

This study was found to have 46% of the CNS anomalies followed by cardiac malformation (23%), Genitourinary system anomalies (10%), Cystic hygroma (5%), 3% each of

Single umbilical artery & Gastrointestinal anomalies & Lung defects etc.

Maternal Age

This study was found to have maximum number of anomalies in the maternal age groups (26-30) years followed by (21-25) years with CNS anomalies commoner in both the groups and CVS anomalies higher prevalence in former age group.

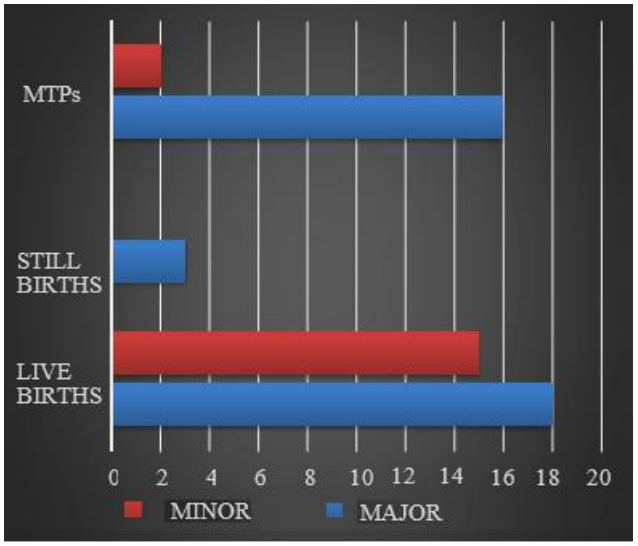

Figure 10: Horizontal Bar graph of Major and Minor anomalies Association

Our study has shown to have 60% of the mothers having live births and 32% undergone MTP.

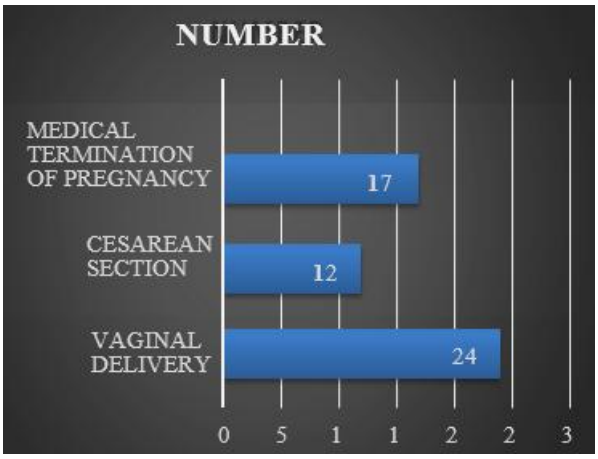

Figure 11: Horizontal Bar graph of Outcome of pregnancies

Our study has shown 45.3% mothers undergone Vaginal deliveries and 22.7% Cesarean section and 32% undergone MTP.

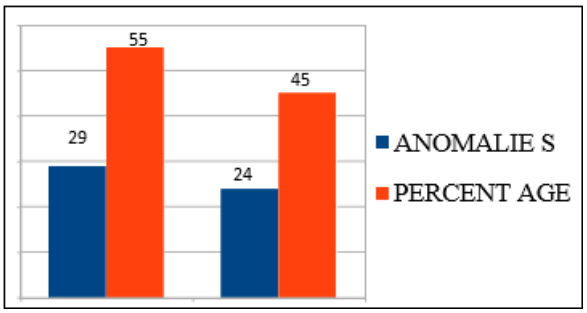

Figure 12: Graph of Sex based Prevalence

Our study had male preponderance with 55% compared to the female fetuses (45%).

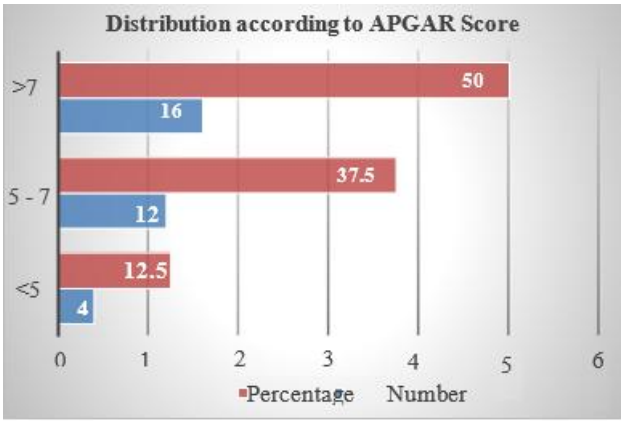

Figure 13: Distribution of newborns according to APGAR Score

In our study 4 babies were born with APGAR Score < 5 and were resuscitated and shifted to NICU but died within 7 days of life. 12 babies were born with APGAR Score 5-7 were shifted to Transient care unit, of which 6 died within 7 days of birth with the remaining requiring surgical procedures. Babies born with APGAR Score > 7 were observed in NICU which eventually had a good prognosis and were discharged with regular follow ups.

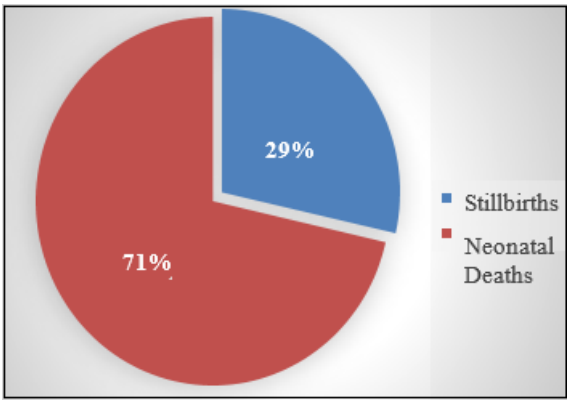

Figure 14: Graph of perinatal mortality

Perinatal mortality rate (PNMR)

Fetal deaths (> 28 weeks of gestation) + Neonatal deaths (< 7 days) Total fetal deaths + Live births (in the same period) x 1000

Our Study found to have 3.5 perinatal deaths per 1000 births; the contribution of stillbirths being 29% and Neonatal

deaths being 71%. Our study showed no maternal morbidity or mortality during the delivery or abortion.

5. Discussion

This study was performed to assess the prevalence of various congenital anomalies and their outcomes in a tertiary care centre in Mumbai.

During this 18 months prospective study in Obstetric department of Tertiary Referral Centre; 53 pregnancies with 61 congenital anomalies were identified among the total births 3900 (including Live / stillbirths) and 132 MTPs.

In our study the prevalence was found to be 1.3% (births and MTP combined) as compared to the prevalence at Kolkata[14] 2.22%; Ahmedabad[5] 2.3%; Pune 1.2% etc and few mentioned in the table below.

Author	Location	Study Period	Prevalence	Male Female ratio
Present study	Mumbai	2020-21 (18 months)	1.3%	1.2:1
Cherian[85]	Vellore	2003-13 (10 years)	1.25%	1.05:1
Parmar[86]	Gujarat	2006-07 (18 months)	0.88%	0.6:1
Egbe[75]	USA	2008 (1 year)	2.8%	1.07:1
Francine[87]	Lebanon	2009 (9 months)	2.4%	1.04:1
Sarkar[16]	Kolkata	2012 (1 year)	2.22%	1.9:1
Malhotra[88]	Punjab	2012 (1 year)	6.8%	5.5:1
Shirke[88]	Dehradun	2012-14 (2 years)	8.39%	1.6:1
Jayashree[90]	Kerala	2009-15 (7 years)	0.84%	1.6:1
Farkhanda[91]	Rawalpindi	2016 (1 year)	0.52%	1.5:1

The above table gives the prevalence of congenital anomalies among various studies and male:female ratio of occurrences. Our study had similar prevalence like the studies done by Baruah[92] et al (1.13%); Patel[93] et al (1.27%); Rao[5] et al (1.21%); Kolah[11] et al (1.4%); Jaikrishan[94] et al (1.4%); Datta and Chaturvedi[95] (1.24%) in India.

Most studies done outside India showed higher prevalence compared to our study, like in Francine[87] et al (2.4%); Egbe et al (2.8%) etc and most of the studies mentioned had considered single tertiary care centre.

In India various studies have also shown a higher prevalence like Malhotra et al (6.8%); Sarkar et al (2.22%); Shirke et al (8.39%); Vishal M Sharma[96] et al (2.48); Yadav[97] et al (2.15%) etc compared to our study probably due to varied definitions of congenital anomalies, period of assessment, varied risk factors across populations. There is also a significant variation in government health care centre and private health care set up.

Region wise variation of the prevalence could be explained by factors like ethnicity, consanguineous marriages, nutritional status of mothers, healthcare utilization patterns.

In our study the proportion of males with congenital anomalies to females was 1.2:1 compared to the studies by Sarkar et al, Shirke et al, Malhotra et al and Jayashree et al etc (ratios being higher); but few studies done by Parmar et al, have showed female preponderance of congenital anomalies; Few studies by Egbe et al, Francine et al, Cherian et al have shown near equal occurrence among male and female fetuses.

In this study the major contribution of the anomalies was among the mothers aged between (25-30 years) being (47%) followed by (32%) and (17%) in the age groups of (21-25years) and (31-35) years respectively which is similar to the studies by Jayashree et al (78% - 21-30 years age); Ghanghoriya et al (82.8% - 21-30 years age); But few studies by Taksande et al; Yadav et al; Hassan Kumarachar et al; Vishal Sharma et al; showed the major proportion of anomalies between the age group of (21-25years) being 72%, 52%, 60%, and (28% - >30 years) respectively.

All fathers were self employed like manual labourer, drivers, security guards etc while most of the mothers were homemakers with 75% of the families belonging to Class-3 (Lower middle class) of the Modified Kuppuswamy Classification. All the patients were from the urban set up residing around the tertiary referral centre and belonged to either Hindu or Islamic religion.

Studies done in Assam (India) by Baruah and Dutta et al and Britain by Sheridan[103] et al and Maria-Morales-Suarez-Varela[104] et al showed significant association of literacy levels and Economic status being protective against developing congenital anomalies directly or indirectly.

In this study we observed that 88.7% of the mothers were in the Normal range of Body Mass Index (18.5 - 24.9) kg/sq.m and only 1 case being overweight (1.9%) similar to study by Dutta et al (71% cases in Normal range and No cases with BMI>23) compared to studies Rankin et al (16.6% cases in the overweight and 15.4% cases in the Obese category); Burgos[108] et al (31% cases with BMI > 25) ; Cai[17] et al, Rasmussen[17] et al and Harris[17] et al showing significant association of Congenital anomalies in relation to Obesity.

In our study we had 1 parent with consanguineous marriage out of 53 pregnancies, suggesting a lesser significance of it with the prevalence of congenital anomalies as compared to studies by Naveed[106] et al (suggesting Odds Ratio of 2.23, p-value 0.01); Hamid[107] et al (52.7% occurrence of multiple anomalies); Sheridan et al ; Stoll et al etc which showed significant association.

In our study we found only 1 mother with a positive family history of congenital anomaly nearly similar to the study by Kumarachar et al and Arjun Singh et al having No/ One family history of anomalies compared to studies by Paladini et al and Kutuk et al which showed significant association for occurrence of congenital anomalies.

Our study had only 1.9% (1) mother with previous history of anomalous birth compared to the study by Ghanghoriya et al having 6.25% of the mothers having previous history.

The prenatal folate supplementation was received in (77.4%) of patients as compared to 61% Shaw et al. The literature provided by Safi et al; Czeizel et al; Werler[105] et al; Wilson et al; Shaw et al etc suggest that the folic acid and multivitamin supplementation provide significant protection from occurring of these anomalies, especially CNS and Spinal anomalies;

It is advisable for all the women of child bearing age group to start folic acid and vitamin supplementation prior to conception. Wilson et al provided a decision tree for folic acid supplementation for prevention of Neural tube defects.

As mentioned earlier, according to CDC[8], classifying congenital anomalies into major and minor categories helps in planning management. We found (64%) of major anomalies in our study as compared to studies by Katherine et al and Ndibizza et al which showed (2.4% and 2% Prevalence) results respectively, indicating the severity of the anomalies.

We observed 4 still births in our study 3 which were associated with major congenital anomalies, as compared to 4.4% of overall deaths due to congenital anomalies at birth as per the Indian consensus of 2020-21 data.

Our study had (47.5%) percentage cases of CNS anomalies as compared to the studies by Ofori et al (1.18%) ; Parker[98] et al (0.02%) ; Ndibazza[99] et al (2%) ; Kumar V[15] et al (13%); Malhotra et al (19%); Taksande et al (8.3%) ; Yadav et al (53%); Macintosh et al (5.6%); Hassan kumarachar[100] et al (74%) ; (84%) Vishal M Sharma[96] et al ; (56%) Ghanghoriya[78] et al ; (21%) Jayashree et al etc. The commonest CNS anomalies detected were Arnold Chiari malformation (17%) and Anencephaly (14%) in our study. Hydrocephalus was most commonly seen in studies by Jayashree et al and Koumi[101] et al and Anencephaly in Vishal M sharma et al. All the anomalies had been detected by Ultrasound scans prenatally.

Our study observed 23% of anomalies being Cardiovascular defects compared to studies showing (19.3%) Saxena HMK et al; (4%) Arjun Singh[102] et al; (12.8%) Hassan

Kumarachar et al; (9%) Mohamed Koumi et al ; (3%) Ghanghoriya et al ; (10%) Yadav et al; (10%) Jayashree et al etc. Ventricular septal defects being most common in ours compared to Atrial septal defects by Ghanghoriya et al and Mohamed Koumi et al.

People involved in the management of fetuses affected with cardiovascular anomalies, e.g. obstetricians, paediatrician cardiologists, and pediatric cardiac surgeons, should be aware of most prognostic data in the literature refer to postnatal series, and the prognosis explained to the parents should be drawn from prenatally detected studies.

Our study had only 1 case of Club foot and that too in association with Iniencephaly comparable to study by Malhotra et al but in contrast to study by Vishal Sharma et al having 10% incidence.

Our study observed about 9.8% of the genitourinary system anomalies in contrast to studies by Koumi et al (13%), Jayashree et al (32%), Yadav et al (10.6%), Ghanghoriya et al (11%), Hassan Kumarachar et al (8.5%) with respective percentage occurrence.

Outcome of Congenital Anomalous fetuses

Our study had perinatal mortality rate of 3.5 deaths / 1000 births compared 1.27/1000 births in EUROCAT registry, extended perinatal mortality rate of 1.3/1000 births in study by Neasham et al, 23/1000 births in study by Cherian et al. This protocol of early detection has led to proper and effective decision making of medical termination of pregnancy in whichever cases indicated, helping the better pregnancy outcomes and reducing the socioeconomic burden over the parents.

In one retrospective study by Zimmer[109] et al in 1997, over a 5year period, with a relatively high detection rate for foetal malformations continuing significant improvement was recorded in the sensitivity of ultrasound examinations (from 53% in the first year to 79% at the end of the study). This resulted in a trend toward more pregnancy terminations and fewer newborns with anomalies over the years.

Our study helps to know the prevalence and patterns of congenital anomalies occurring at a tertiary centre in this part of the country.

The results in our study help to know the significant / insignificant associations of various factors for causation of congenital malformations. Most of the observations are comparable with the similar studies undertaken in other parts of the country. However, some of the observations differ which is expected given the different nature of various studies like hospital versus community based, the sociodemographic and ethnic variations, difference in geographical and environmental factors, difference in time period for follow up, criteria for defining and management plan for specific anomalies.

- 45% of the babies were delivered vaginally and 22.7% by cesarean section, 32% have undergone MTP.
- Our study had fetuses with male preponderance.
- Out of 32 live births, 10 newborns died within first 7 days of life, 8 newborns advised surgical management and remaining were discharged with routine follow up and care.
- The main purpose of our study was to involve and make aware all the health care workers who are providing maternal and child health care working in government or private sector so as to quantify exact prevalence rate of congenital malformations involving any particular system.
- Our study definitely helps to know the pattern of congenital anomalies in this region of the country so that strategies for prevention, early detection and timely management can be sort out.

6. Conclusions

Congenital anomalies are important cause of perinatal mortality and morbidity, most of which are preventable, causes being infections, addiction and nutrition. Non preventable causes like genetic disorders, congenital anomalies have greater importance as a significant cause of morbidity and mortality in all stages of life. This includes incidence or prevalence of these disorders, associated morbidity and mortality, life expectancy and socioeconomic burden on the family and society. In our study it was found that overall prevalence of congenital anomalies was 1.3% (including live / stillbirth / Medical Termination). Central nervous system anomalies were most commonly detected anomalies.

Awareness of antenatal, natal and postnatal care has decreased perinatal mortality in general especially those caused by anoxia and infection, however perinatal deaths due to congenital malformation still remains as a major group. Congenital malformations due to preventable causes like alcohol, infections, folic acid deficiencies can be reduced by spreading awareness or educating women of child bearing age group through preconceptional counselling.

Chromosomal abnormalities leading to developmental anomalies if diagnosed at an early gestational age can be terminated. Chromosomal abnormality which do not cause structural malformation, but cause neonatal metabolic disorders are difficult to diagnose clinically unless family history or history of metabolic disorder in previous child is available. Invasive prenatal testing with genetic screening can be carried out in such cases.

The mortality and morbidity of the anomalous foetuses or infants is due to lack of resources and lack of availability of invasive procedures and genetic lab at peripheral hospitals. Our study showed 32% of medical termination of pregnancies due to effective and appropriate antenatal ultrasound screening available at our tertiary care centre with experienced staff.

Further studies are required with good reliable diagnostic facilities to reduce the etiological factors involved in causation of malformations. This helps in predicting the future recurrences so as to undertake prenatal genetic counselling and to prevent fatal congenital malformations.

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