Section: Paediatrics

Original Research Article

PATTERN OF CONGENITAL MALFORMATION IN NEWBORN AND THEIR SHORT TERM OUTCOMES IN TERTIARY REFERRAL CENTRE

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Abstract

Background: Congenital malformation is functional or structural anamolies identified prenatally, at birth, or may be later in life. They contribute to about 10 percent of neonatal deaths. Based on WHO report about 3 million fetuses and infants are born each year with major congenital malformation. The incidence of major congenital malformation is 2-3 % of all live births with more than 60% identified in first month. The incidence of major congenital malformation is even higher in still births. The Primary aim is to determine the prevalence of major and minor congenital malformations among all neonates admitted in a tertiary referral centre. The secondary aim is follow up of all newborns with malformations to delineate the mortality and morbidity profile. **Materials and Methods:** This was a descriptive observational study done from march 2019-february 2020 at SNCU Coimbatore medical college with 3 months follow up .The study included all live newborns admitted in SNCU with exclusion of still births . Ethical committee clearance was also obtained from the Institutional Ethics Committee of the hospital Informed and written consent were obtained from parents after explaining in detail about the method of the study and the procedures involved Major and minor congenital malformation were classified system wise according to ICD1026. **Result:** About 4200 babies were born during the period of which 494 had congenital malformations with 452 major and 42 minor respectively. Of the 494 babies term with male gender contributed to 63%.Major congenital malformation was with CVS anamoly contributing to26% followed by CNS and GIT. Minor malformations contribute to 9% of the anamolies with preauricular tag present in 17% of newborn babies. On 3 month follow up of babies 222 babies were managed medically, 202 had surgical correction with 70 babies losing their life in early neonatal period. **Conclusion:** The key findings from the study suggest that the first requirement for a birth defects policy is surveillance of birth defects to help the description of the epidemiology and public health impact of congenital anomalies, and anticipate the health care needs for birth defects. Surveillance should also provide data on the number of infants with special needs, so as to provide support for childhood disability and children with chronic medical needs.

INTRODUCTION

Congenital malformations are morphologic defects that originate in the prenatal period as result of genetic mutations, chromosomal aberrations and/or adverse intrauterine environment, that contribute to approximately 4% to 6% of newborns with major birth defect and require genetic evaluation.[1,2]
Congenital anomalies are the fifth largest cause of neonatal mortality contributing approximately to 10% of neonatal deaths.Identification of the cause for anamolies would be helpful in reducing the incidence, treating them ,preventing these in future pregnancies, and proper planning of subsequent pregnancy with proper genetic counseling.[3-5]
These anomalies are functional or structural anomalies that can be identified prenatally, at birth, or may be only diagnosed later in life as in cases with hearing defects. The etiology of up to 60% of congenital anomalies is still unknown, with 6-10% chromosomal, 3-7.5% single gene Mendelian disorders, 20-25% multi factorial and 4-5% case due

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environmental exposures. The common reported risk factors include consanguinity, nutritional deficiency, infections and teratogenic drugs. Studies show that 20-40 per 1000 live births of newborn are born with congenital anamolies.[6-10]

Based on a World Health Organization (WHO) report, about 3 million fetuses and infants are born each year with major congenital malformations; which accounts for an estimated 495,000 deaths worldwide. Of all the congenital malformations diagnosed nearly60% are identified in the first month and about 80% by the end of 3 months. The prevalence of major congenital malformations is even higher among stillbirths with a significant birth defect reported in 15–20%. With the introduction of prenatal ultrasound the termination of pregnangy due to congenital malformation has risen to 47%from 23% between 1985-2000 25. diagnostic accuracy for detecting anencephaly by prenatal ultrasound is 90% and for gastrointestinal anamolies is less than 70% like diaphragmatic hernia, bladder outlet obstruction, and major skeletal defects. Similarly, many cardiac defects diagnosed in the first year of life are not detected prenataly. Studies from Europe, Canada have shown increase in prenatal diagnosis of major malformation and increase in termination of pregnancy. Studies from UK reported increase in congenital anamolies of nonchromosomal origin with increasing socioeconomic deprivation.[11-20]

Shivanagappa et al., conducted a retrospective study of the incidence of congenital anomalies and their maternal risk factors of which history of previous abortions (27.7%), consanguinity (10.6%) and anomalous uterus (6.4%) were significantly identified as risk factor.[21]

According to Jenita Baruah et al,[2] a cross sectional study was undertaken from May 2010 through Feb 2013 for estimation of gross congenital malformations in Assam Medical College. A total of 18,192 births including live births and still births were examined of which 206 cases of structural malformations were observed. Prevalence of congenital malformations was 1.2%. of total live births. Distribution of malformations was predominant among males than in females. (60.67% vs 37.37% $p<0.005$). Musculoskeletal system was the most common system involved with maternal age above 30 yrs as important risk factor.[22-25]

A cross sectional study conducted by Shrestha et al, at Kathmandu Medical College showed that, out of 2456 live births examined, congenital malformations were observed in 66 cases with the prevalence of 66 (2.6%) at 95% confidence interval (4.19-1.98) of total live births. The genitourinary system was the most common system involved with congenital malformations being 16 (24.2%), followed by musculoskeletal system 14 (21.2%), and cardiovascular system (18.2%).[26-28]

Sozan K. Ameen, Shahla Kareem Alalaf, and Nazar P. Shabila presented an analysis of anomalies observed in 35,803 recorded births in the Maternity Hospital in Iraqof which 130 babies had congenital anomalies, giving a rate of 36.3/1000 deliveries. The most common areas for anomaly was the central nervous sytem (37.7%) followed by musculoskeletal system (23.1%) and gastrointestinalsystem (20.8%). There was a statistically significant association between having a child with congenital anomalies and maternal history of previous congenital anomalies.

A retrospective study by Kishan Chand Gupta at a hospital in Uttar Pradesh, a total of 3450 live births were studied for congenital malformation. There were 106 malformed babies found with prevalence of 31.8%. Congenital malformations of the central nervous system were the highest followed by those of musculoskeletal system, gastrointestinal system, cardiovascular system, genitourinary system, chromosomal and ear.

Aim of the Study

- To determine the prevalence of major and minor congenital malformations among all neonates admitted in a tertiary referral centre.
- Follow up of all newborns with malformations to delineate the mortality and morbidity profile.

Justification of the Study

- Estimation of congenital malformations and their associated risk factors will be helpful in policy making and prevention strategy at a higher level
- Identifying the congenital malformations and early intervention through District Early Intervention Centre may improve the overall outcome of the babies.

MATERIALS AND METHODS

Study design: Descriptive study

Study Period: March 2019 to February 2020 and 3 months follow up.

Study Place: Coimbatore Medical College Hospital Sick Newborn Care Unit.

Study population: All newborns admitted in NICU

Inclusion criteria:

All newborns admiited incmchnicu

Exclusion criteria: Stillborn babies

Consent and ethical clearance: Informed and written consent were obtained from parents after explaining in detail about the method of the study and the procedures involved. Ethical committee clearance was also obtained from the Institutional Ethics Committee of the hospital.

All the mothers were interrogated within 48 hours of delivery as per the proforma prepared that contained the particulars like maternal age, consanguinity, education, socioeconomic status, and antenatal history in detail with reference to drug intake, fever, and exposure to irradiation. Medical diseases complicating pregnancy like Diabetes, Heartdisease, and hypertension were also taken into account. A detailed obstetric history with reference to previous abortions and stillbirths was taken from the mother. Information was obtained on fetal activity, size and position. Perinatal information including gestational age, fetal position at delivery, duration of labor, type

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of delivery and any evidence of fetal distress was obtained.

A comprehensive family history comprising of three-generation pedigree was elicited. Reproductive history especially about infertility, miscarriages, and stillbirth were inquired. A complete physical examination of all new born including measurements of length, weight, and head circumference were taken and compared with standard charts. The shape and size of the head and fontanelle as well as the cranial sutures were noted, with assessment for evidence of craniosynostosis or an underlying brain malformation. Spacing of the eyes and presence or absence of coloboma were noted.

Ears were examined for presence of pre auricular tags, sinuses, pits or abnormal crease, along with placement, length and folding. Evaluation of nose, the shape of nasal tip, the alar nasi, presence of anteverted nares, patency of choanae were done. The mouth and throat were examined for the presence of cleft lip and palate; the shape of palate and uvula were noted, and the presences of unusual features like natal teeth were recorded. Retrognathia or receding chin, which could be a part of a syndrome, was also noted. Neck was inspected for nuchal folds or webbing. Evaluation of chest and thorax including lung auscultation and cardiac examination was done. Any obvious thoracic deformity was noted. Abdominal examination was focused on determining any defect in anterior abdominal wall like exomphalos, sdeficient anterior abdominal wall muscles. Presence of organomegaly was documented. Umbilicus was also examined for hernia as well as number of vessels present in cord.

Genitourinary examination was concentrated on determining whether anomalies such as hypospadiasis, cryptorchidism, microphallus and ambiguous genitalia were present. The anus was examined for evidence of tags, its placement and patency. The back was also examined especially for the shape of the spine, any associated defects such as meningomyelocele, natal cleft, dermal sinus sacral dimple

Hands and feet were assessed for polydactyly, syndactyly, and clinodactyly, club foot. Examination of skin for phakomatoses like café aulait macules, hemangiomas, port-winestains was done.

Radiographs, ultrasonogram, and echocardiogram were done in babies for selective cases. The anomalies were grouped and categorized as syndrome, sequence, association or field defect. They were also classified as major, minor, or normal variant. All the datas were tabulated and analyzed statistically.

Major and minor classicfication was done by using ICD10 classification (26): results were collected and analysed.

RESULTS

A total of 4200 babies were admitted over a period of 1 year from March 2019 to February 2020 at Coimbatore Medical College Hospital SNCU. A total of 494 babies with congenital malformations were admitted. Among them, 452 babies were sadmitted for major congenital anomalies and 42 babies were admitted with minor malformations.

All the results were tabulated and analysed statistically. Demographic and qualitative variables in the qualitative form were expressed as frequencies with their percentages.

Association between type of anomalies with demographic/clinical variables variables were analysed using Pearson chi- square.

Findings

Among the malformed babies,180babies were female (36.4%),314 were males contributing to 63.6% of total cases. Approximately 64% of congenital malformations occurred in male babies compared to female babies.

Mothers were classified according to their age into 3 groups. The incidence of malformed babies were found to be common in age group of 20-30 years. Of 494 malformed babies, 424 babies were born to mothers of age group 20-30 years.

It was observed that, there is an increase in incidence of congenital anomaliesin multi gravida mothers. Approximately, 66% congenital malformed babies were born to multi gravida mother.

Of 494 babies with congenital anomalies,284 babies were delivered by LSCS Term babies with congenital malformations constituted 84.8% whereas preterm babies constituted 15.2% of the total.

Of the 494 malformed babies, 83.6% babies were born of non consanguineous marriage as compared to 16.4% babies who were born of consanguineous marriage.

49.8 % of anomalies were antenatally diagnosed whereas the remaining50.2% were not diagnosed by antenatal scans.

Most of the anomalous babies were in the weight group of 2-3 kg.

On evaluation of maternal complications during pregnancy, 342 mothers had antenatal complications and 152 mothers had illness. Out of 152 mothers, 77 had Pregnancy Induced Hypertension, 49 had Gestational Diabetes Mellitus and rest had complications like anaemia or hypothyroidism complicating pregnancy.

The commonest system involved in anamoly was the cardiovascular system(129 cases), followed by central nervous system (82 cases), GIT(77 cases), genito urinary system (69 cases).

Major anomalies of cardiovascular system were ventricular septal defect followed by transposition of great vessels. Among the CNS malformations, meningomyelocele was the most common anomaly, accounting for 46.3% of CNS malformations, followed by congenital hydrocephalus.

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Among the congenital malformations of gastro intestinal system, congenital diaphragmatic hernia (40.3%), was the most common anomaly followed by duodenal atresia (18.2%).

Out of 494 babies, 69 had genito urinary anomalies, of that 42% (29 cases) was contributed by hydroureteronephrosis. This was followed by posterior urethral valve and ectopic kidney.

A total of 15 syndromic babies were admitted during the study period. Of that, the most common was Down syndrome (46.7%) followed by Pierre Robin sequence (40%).

During the study period, 12 babies with cutaneous malformations were admitted.5 had aplasia cutis and another 5 were collodion babies. Cutaneous malformations contributed around 2.4

Minor malformations contribute around 9% of total babies admitted with congenital malformations. The most common minor anomaly observed was pre auricular tag followed by single umbilical artery.

A 3 month follow up was done for all babies born with congenital anomalies. Among them, 70 babies lost their lives either during their SNCU stay or in the early neonatal period. Rest of them were followed up. Out of them, 222 babies were managed medically and 202 babies were managed with appropriate surgery.

Out of 70 deaths, 44% was constituted by gastrointestinal anomalies out of which 61% was constituted by congenital diaphragmatic hernia followed by tracheo-oesophageal fistula.

Next most common cause of mortality occurred in babies with anomalies of the cardiovascular system of 34%. Total anomalous pulmonary venous connection was the most common cause of death in cardiovascular malformations (8 cases) followed by transposition of great arteries (6 cases).

In central nervous system, meningomyelocele was the leading cause of death i.e. 8 cases (11%).

This study has found that there is a statistically significant association between PIH and gastro intestinal anomalies (p value<0.005).

There was also a statistically significant association between PIH and genitor urinary anomalies (p value< 0.005).

There was a statistically significant association between gestational diabetes mellitus and cardiovascular anomalies (p value <0.001).

Maternal age more than 30 years increased the risk of occurrence of Down syndrome. In this study, 5 out of 7 mothers of Down syndrome were above 30 years of age which has a statistically significant p value < 0.001.

Maternal folate intake during peri conceptional period was found to reduce the risk of meningomyelocele. In the present study, 40% of mothers have not consumed folic acid in pre conceptional period. Folic acid was started only after confirmation of pregnancy.

There was a statistically significant association between polyhydramnios and gastro intestinal system anomalies especially tracheo esophageal fistula, duodenal atresia and jejunal atresia. (P value was significant at <0.001 level).

Table 1: Sex Distribution and Congenital Malformations.

Gender	Frequency	Percentage
Male	314	63.6
Female	180	36.4
Total	494	100.0

Table 2: Distribution Of Malformations According to Maternal Age

Maternalage	Frequency	Percentage
<20	53	10.7
20-30	424	85.8
>30	17	3.4
Total	494	100.0

Table 3: Distribution According To Parity

Parity	Frequency	Percentage
Primigravida	327	66.2
Multigravida	167	33.8
Total	494	100.0

Table 4: Mode Of Delivery

Mode of delivery	Frequency	Percentage
Nvd	210	42.5
Lscs	284	57.5
Total	494	100.0

Table5: Distribution According To Gestational Age:

Gestationalage	Frequency	Percentage
Term	419	84.8
Preterm	75	15.2
Total	494	100.0

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Table6: Consanguinity And Congenital Malformations:

Consanguinity	Frequency	Percentage
Consanguinous	81	16.4
Nonconsanguinous	413	83.6
Total	494	100.0

Table 7: Distribution of Cases According To Birthweight

Weight	Frequency	Percentage
<2kg	83	16.8
2-3 Kg	405	82
>3kg	6	1.2
Total	494	100.0

Table8: Congenital Malformations and Antenatalscans

Andiagnosed	Frequency	Percentage
Yes	246	49.8
No	248	50.2
Total	494	100.0

Table9: Complications during Pregnancy and Congenital Malformations

Ancomplications	Frequency	Percentage
Pih	77	15.6
Gdm	49	9.9
Hypothyroidism	10	2.0
Anaemia	9	1.8
Polyhydramnios	7	1.4
Nil	342	69.2
Total	494	100.0

Table10: Systemwise Distribution of Congenital Malformations

System wise anomalies	Frequency	Percentage
Cvs	129	26.1
Cns	82	16.6
Git	77	15.6
Musculoskeletal	25	5.1
Syndromes	15	3.0
Cleftlip&Cleftpalate	40	8.1
Genito-Urinary	69	14.0
Skin	12	2.4
Miscellaneous	45	9.1
Total	494	100.0

Table 11: Cardiovascular System Analysis

Cvsanomalies	Frequency	Percentage
Vsd	46	35.7
Tga	22	17.1
Tapvc	13	10.1
Asd	8	6.2
Pda	8	6.2
Tof	10	7.8
Coarctationofaorta	5	3.9
Hypoplasticltheartsyndrome	4	3.1
Pulmonaryatresia	4	3.1
Situsinversustotalis	3	2.3
Truncusarteriosus	3	2.3
Ebsteinanomaly	1	0.8
Hypoplasticltheartsyndrome	1	0.8
Tricuspidatresia	1	0.8
Total	129	100.0

Table12: Analysis Of Centralnervous System Malformations

Cnsanomalies	Frequency	Percentage
Meningomyelocele	38	46.3
Congenitalhydrocephalus	21	25.6
Choroidplexuscyst	10	12.2
Arnoldchiarimalformation	6	7.3
Corpuscallosalagenesis	3	3.7
Craniosynostosis	3	3.7
Holoprosencephaly	1	1.2

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Total	82	100.0

Table13: Analysis of Gastro Intestinal System

Gitanomalies	Frequency	Percentage
Cdh	31	40.3
Duodenalatresia	14	18.2
Tef	10	13.0
Anorectalanomaly	13	16.9
Jejunalatresia	6	7.8
Gastroschisis	2	2.6
Illealatresia	1	1.3
Total	77	100.0

Table14: Analysis Of Genitourinary System:

Genito-Urinary	Frequency	Percentage
Hun	29	42
Puv	8	11.6
Ectopickidney	6	8.7
Kidneyagenesis	6	8.7
Exostrophybladder	5	7.2
Hypospadias	4	5.8
Dysplastickidney	4	5.8
Mcdk	3	4.3
Hydrocele	2	2.9
Pujobstruction	2	2.9
Total	69	100.0

Table 15: Chromosomal Disorders and Other Syndromes

Syndromes	Frequency	Percentage
Pierrerobinsequence	6	40
Downsyndrome	7	46.7
Edwardsyndrome	1	6.7
Patausyndrome	1	6.7
Total	15	100.0

Table 16 Cutaneous Malformations

Skin	Frequency	Percentage
Aplasiacutis	5	41.7
Collodionbaby	5	41.7
Ectodermaldysplasia	2	16.7
Total	12	100.0

Table17: Minor Malformations

Miscellaneous	Frequency	Percentage
Singleumbilicalartery	7	15.0
Umbilicalhernia	6	13.3
Preauriculartag	8	17.8
Natalteeth	4	8.9
Polydactyly	4	8.9
Tonguetie	4	8.9
Preauricularsinus	3	6.0
Syndactyly	3	6.0
Microphthalmia	2	4.4
Micrognathia	1	2.2
Total	42	100.0

Table 18: Outcome of the Study

Medical	222
Surgical	202
Death	70

Table 19: Systemwise Analysis of Death

Cardiovascularsystem	24
Gastrointestinalsystem	31
Centralnervoussystem	8
Genitourinarysystem	2
Miscellaneous	5

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Table20: Association Between Maternal Illness And Incidence Of Anomalies:Pih And Congenital Anomalies

System		Pih				Chisquaretest P Value
		Yes		No		
		N	%	N	%	
Cvs	Yes	13	16.9%	116	27.8%	0.045
	No	64	83.1%	301	72.2%	
Cns	Yes	11	14.3%	71	17.0%	0.553
	No	66	85.7%	346	83.0%	
Git	Yes	22	28.6%	55	13.2%	0.001*
	No	55	71.4%	362	86.8%	
Musculo Skeletal	Yes	2	2.6%	23	5.5%	0.283
	No	75	97.4%	394	94.5%	
Syndromes	Yes	2	2.6%	13	3.1%	0.807
	No	75	97.4%	404	96.9%	
Cleftlip& Cleftpalate	Yes	6	7.8%	34	8.2%	0.915
	No	71	92.2%	383	91.8%	
Genitourinary	Yes	19	24.7%	50	12.0%	0.003*
	No	58	75.3%	367	88.0%	
Skin	Yes	0	0.0%	12	2.9%	0.132
	No	77	100.0%	405	97.1%	
Miscellaneous	Yes	2	2.6%	43	10.3%	0.031
	No	75	97.4%	374	89.7%	

Table21: Gdm and Congenital Anomalies

System		Gdm				Chi square test P-value
		Yes		No		
		N	%	N	%	
Cvs	Yes	35	71.4%	94	21.1%	<0.001*
	No	14	28.6%	351	78.9%	
Cns	Yes	5	10.2%	77	17.3%	0.205
	No	44	89.8%	368	82.7%	
Git	Yes	3	6.1%	74	16.6%	0.054
	No	46	93.9%	371	83.4%	
Musculo Skeletal	Yes	1	2.0%	24	5.4%	0.310
	No	48	98.0%	421	94.6%	
Syndromes	Yes	1	2.0%	14	3.1%	0.669
	No	48	98.0%	431	96.9%	
Cleft Lip & Cleftpalate	Yes	0	0.0%	40	9.0%	0.029
	No	49	100.0%	405	91.0%	
Genitourinary	Yes	1	2.0%	68	15.3%	0.011
	No	48	98.0%	377	84.7%	
Skin	Yes	0	0.0%	12	2.7%	0.245
	No	49	100.0%	433	97.3%	
Miscellaneous	Yes	3	6.1%	42	9.4%	0.444
	No	46	93.9%	403	90.6%	

Table22: Maternal Age and Down Syndrome

		Down syndrome				Chisquaretest P Value
		Yes		No		
		N	%	N	%	
Maternal Age	<20	0	0.0%	53	10.9%	<0.001*
	20-30	2	28.6%	422	86.7%	
	>30	5	71.4%	12	2.5%	
	Total	7	100.0%	487	100.0%	

Table23: Maternal Folate Intake and Meningomyelocele

		Meningomyelocele				Chisquaretest P Value
		Yes		No		
		N	%	N	%	
Folateintake	Yes	30	83.3%	434	94.8%	0.006
	No	6	16.7%	24	5.2%	
	Total	36	100.0%	458	100.0%	

Table24: Significance of Polyhydramnios In Antenatal Scans

System		Polyhydramnios				Chisquaretest P Value
		Yes		No		
		N	%	N	%	
Cvs	Yes	0	0.0%	129	26.5%	0.113
	No	7	100.0%	358	73.5%	
Cns	Yes	0	0.0%	82	16.8%	0.235

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	No	7	100.0%	405	83.2%	
Git	Yes	7	100.0%	70	14.4%	<0.001*
	No	0	0.0%	417	85.6%	
Musculo Skeletal	Yes	0	0.0%	25	5.1%	0.538
	No	7	100.0%	462	94.9%	
Syndromes	Yes	0	0.0%	15	3.1%	0.637
	No	7	100.0%	472	96.9%	
Cleft Lip & Cleftpalate	Yes	0	0.0%	40	8.2%	0.429
	No	7	100.0%	447	91.8%	
Genitourinary	Yes	0	0.0%	69	14.2%	0.283
	No	7	100.0%	418	85.8%	
Skin	Yes	0	0.0%	12	2.5%	0.674
			100.0%	475		
Miscellane Ous	Yes	0	0.0%	45	9.2%	0.399
	No	7	100.0%	442	90.8%	

DISCUSSION

Congenital anomalies are not prioritized as public health problems in low income countries as they are considered to be rare conditions that are self- limiting due to the high mortality of affected infants. Another reason for under-prioritization of the condition is the understanding that most birth defects are not preventable through low cost primary care strategies. Neural tube defects like anencephaly could be prevented potentially through a low cost prevention method of periconceptional folic acid supplementation. When combined with pre conception iron supplementation, this primary care intervention might not only reduce the number of neural tube defects, but also anaemia, which is a persistent maternal health challenge in India.

Prevalence

In the present study, among 4200 admissions in SNCU during the study period, 494 babies were admitted for congenital malformations. The frequency of congenital malformations in the present study was 11.7%. (494 cases). Among them, major malformations contributed to 452 cases around 91% of total cases, minor anomalies accounted for 9% i.e. 42 cases.

In a study by Vikram Datta et. (6), out of 2968 deliveries, 2869 were live births and 99 were stillbirths. The number of babies with congenital anomalies was 48. Out of these 26 (70.3%) babies had major anomalies and 11 babies (29.7%) had 14 minor anomalies.

In our study the prevalence was much higher than other studies the cause being antenatal detection of major anamolies

Sex distribution: In a study by Shatanik Sarkar et al,[1] more male babies were born with congenital anomalies than female babies. Among the malformed babies 314 babies were male and 180 were female with Male female ratio was 1.7 which was statistically significant and similar in our study

But in a study conducted by Vikram Datta et al,[6] and Verma et al,[10] there was no difference in the distribution of malformations between the two sexes.

Consanguinity and congenital malformations: According to a study by Rittler M.,et al,[24] a significant association with parental consanguinity was observed for three congenital anomalies: bilateral cleft lip +/- cleft palate, hydrocephalus and postaxial polydactyly. In the present study, 84% babies were born of non consanguineous marriage and 16% babies were born of consanguineous smarriage. It was found that there was no significant association between congenital anomalies and consanguinity.

Maternal age and anomalies: Suguna Bai et al,[11] reported a higher incidence of malformation in the babies born to mothers of age more than 35 years, whereas a study by Dutta et al,[6] showed there was no statistically significant association between increased maternal age and congenital anomalies. In this study, it was revealed that a majority of malformed babies were born to mothers of age group 20-30 years. Also it was found that there was a significant association between increasing maternal age and Down syndrome. However on further analysis, 5 out of 7 Down syndrome babies were found to have been born to mothers above 30 years of age which was statistically significant.

Maternalillness and congenital malformations: In this study around 16% of mothers had Pregnancy Induced Hypertension followed by Gestational Diabetes Mellitus of around 10%. It was found that maternal illness had an impact on incidence of anomalies in babies.

Pregnancy induced hypertension had significant association with gastrointestinal system as well as genitourinary system anomalies. 28.7% of babies GIT anomalies had mothers with PIH and24.7% mothers with PIH gave birth to babies with genitor urinary anomalies On further analysis, in GIT, the most common anomaly associated with PIH was Congenital diaphragmatic hernia. In genitourinary system, the most common anomaly was hydrouretronephrosis.

Maternal GDM had a significant association with cardiovascular anomalies. 35 babies with cardiovascular anomalies were born to GDM mothers which was around 27% of total cardiovascular anomalies and this result was statistically significant.

In this study,there was no significant association between maternal drug intake and incidence of congenital anomalies. There was also no significant relation between other maternal illness like hypothyroidism/ anaemia complicating pregnancy and congenital anomalies

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Significance of antenatal scans: Data on detection rates using ultrasound for screening for fetal malformations do vary widely, showing a range from8.7% to 85%. The overall sensitivity for ultrasonographically detectable fetal malformations was 35% in tertiary facilities which was significantly higher compared to 13% in community hospitals which suggests that operator experience, skills and training are important determinants. Of 494 babies with congenital anomalies, 50% of anomalies were diagnosed antenatally. Antenatal scans were sensitive in detecting cardiovascular, gastrointestinal, central nervous system and genitourinary system anomalies compared to other musculoskeletal and minor anomalies.

This study also found that polyhydramnios was one of the most significant indicators of gastrointestinal system malformations. P value was significant <0.001. Hence possibility of gastrointestinal malformations should always be kept in mind if polyhydramnios is detected in antenatal scans.

Systemic distribution of congenital malformations: Malformations of the cardiovascular system was the most common in the present study followed by central nervous system. However some studies have shown CNS defects,[21] being the highest while one study had reported the most common system involved to be gastrointestinal system.[11]

A study byTaksande et al,[3] showed cardiovascular anomalies to be the most common. Our study conforms to this data.

Health service implications: In this study, several babies with anomalies needed care, in the form of corrective surgery, orthopaedic intervention for CHDs, CTEV, cleft lip/palate, diaphragmatic hernia and tracheo esophageal fistula respectively

So pediatric surgery services for congenital anomalies have to be included as a component of newborn care.

Increasing awareness of maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be emphasized to decrease the incidence of congenital anomalies and their co morbidities.

The second health care implication is the utilization of ultrasonography for the detection of fetal anomalies. It was noted that upon detection of an anomaly, and when there was no option to terminate the pregnancy, mothers were left to continue the pregnancy till term without any psychosocial support or counselling. Thus, the study observed the need for appropriate pre- and post- test counselling and dissemination of knowledge about the utility of ultrasonography in the detection of congential anomalies.

The mortality rate was observed to be high in gastrointestinal anomalies followed by cardiovascular anomalies. Pediatric cardiothoracic surgical facilities are still not accessible and not affordable in all tertiary care centres. So the study emphasizes the need for superspeciality care in tertiary centres to reduce death due to complex congenital heart diseases.

CONCLUSION

The key findings from the study suggest that the first requirement for a birth defects policy is surveillance of birth defects to help the description of the epidemiology and public health impact of congenital anomalies, and anticipate the health care needs for birth defects.

Surveillance should also provide data on the number of infants with special needs, so as to provide support for childhood disability and children with chronic medical needs.

In the present study, it was found that there was significant association between the incidence of gastrointestinal and genitourinary anomalies with maternal hypertension. Further studies should be conducted to analyze the relation of maternal hypertension with GIT and genitourinary anomalies and also analysis should be done as to which anomaly is the most common in that system and the specific outcome.

Further experimental studies like folic acid fortification of flour to reduce the incidence of neural tube defects can be conducted as in western countries Studies regarding this are still lacking in India which could be taken up for future reduction in the incidence of neural tube defects.

Stillborn babies may be included in future studies so as to get the actual prevalence of congenital anomalies

Limitation of the Study

As ours is a tertiary care referral centre, the prevalence calculated may be higher than that in general population. Tertiary care hospitals do not usually have a definite catchment area and complicated cases are more often encountered. Hence the data cannot be projected to the general population. So population based studies are necessary.

Still born babies were not included in the study.

Minor malformations may go unnoticed as it is a study conducted in a referral centre and most commonly major malformations are referred for further care.

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