

Study of clinically manifested congenital malformations in live birth

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Abstract

Congenital malformations being an important cause of neonatal mortality both in developed and developing countries. It is also a major cause for fetal loss, and also contributes significantly to premature birth, childhood and adolescence morbidity. So, this study aims in early detection, treat and take steps against preventable causes in subsequent pregnancies.

Materials and method: The parity and age of mother of malformed newborns will be noted and analysed. The paternal habits of smoking, chewing tobacco and alcohol consumption will also be recorded. The diagnosis of congenital malformations will be based on analysis of antenatal records, clinical examination and further investigations, if required.

Conclusion: Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

Keywords: prenatal, planned termination, malformations, premature birth

1. Introduction

Congenital malformations are morphological defects that occur in the prenatal period as a result of genetic mutation, chromosomal abnormalities and adverse intrauterine environment. These are present at birth and clinically manifest at any time in life [1].

Congenital malformations have been known and recognized for centuries. It is a problem for research because of high frequency of their occurrence and devastating effect they may have on the individual and family. For around 8% to 15% of the perinatal death and 13% to 16% of the neonatal death congenital anomalies are responsible [2, 3].

Congenital malformations cause a noteworthy share of infant mortality and morbidity and health care expenditure. As a consequence, it is essential to have basic epidemiological information of these anomalies. Congenital anomaly rates have also been used for primary health services planning.

The most traumatic experience for a gravid woman, her spouse and the family members, is, undoubtedly, the unexpected birth of deformed child, precipitating feeling of horror, inadequacy and failure in parents [1]. Congenital malformation represents defects in morphogenesis during early fetal life.

Patients with multiple congenital anomalies present are not so frequent but tremendously difficult challenge to the pediatricians. Also, increased use of irradiation, alkylating agents, anti-metabolites, drugging, smoking, alcohol consumption has contributed to a spike in numbers in congenital anomalies. It has been estimated that one in 40 or 2.5% of newborns have are cognizable malformation or malformation at birth.

In India with decreasing mortality due to infection and nutritional disorders, incidences in death due to congenital malformation are increasing [5]. A study done at AIIMS

show that congenital malformations contributed to 13.4% of perinatal deaths as while 10 years back it was 9% [8]. Major malformation accounts for 15% of neonatal death [6].

Congenital malformations being an important cause of neonatal mortality both in developed and developing countries [3, 4]. It is also a major cause for fetal loss, and also contributes significantly to premature birth, childhood and adolescence morbidity.

Hence the present study was done at our tertiary care centre to detect congenital malformations at the earliest, treat correctable causes and take preventive and avoidable steps in the next pregnancy against preventable causes.

Aims and objectives

Aims

- To study the spectrum of clinically manifested congenital malformations among live births at Tertiary care hospital.
- To study the incidence of clinically manifested congenital malformations among live births at Tertiary care hospital

Objectives

Early detection for the clinically manifested congenital malformations and thus helping in preventing similar malformations in further pregnancies.

Materials and Methods

Source of data

Newborn babies who are delivered in, Krishna Hospital, Karad during the study period of October 2016 to March 2018.

Study design

Cross sectional study

Type of Study

Cross-sectional study from 1 October 2016 to 31 March 2018. Method of Collection of Data (including sampling procedure if any): All live births at Krishna Hospital medical college will be studied from October 2016 to March 2018. All the newborns will be examined for visible structural anomalies and required investigation will be done if required.

Inclusion Criteria

All newborns delivered at Krishna hospital will be examined clinically and investigated further, if required.

Exclusion criteria

New born with congenital malformations born outside Krishna Hospital. The functional anomalies (e.g. metabolic disorders) shall be excluded from the study.

Method of examination

The parity and age of mother of malformed newborns will be noted and analysed. The paternal habits of smoking, chewing tobacco and alcohol consumption will also be recorded. The diagnosis of congenital malformations will be based on analysis of antenatal records, clinical examination and further investigations, if required.

- Information will be collected in semi structured proforma in each case with consent of parents.

Observations and results

A cross sectional study in 4786 live births was conducted to study the spectrum and incidence of clinically manifested congenital malformations among live births. It was noted that out of the total 4786 live births, the number of babies with congenital malformations was 100.

Table 1: Percentage of malformed cases in Live Births

Total Live Births	4786
Malformed Cases	100
% of malformed cases in Live Births	2.09%

Gender of Neonates

There was 2753 (57.5%) and 2033 (42.5%) male and female births. Male borns had higher incidence of congenital malformation compared to females (68 vs. 32 cases respectively) as per Chi-Square test ($p < 0.05$).

Table 2: Gender of Neonates

Gender	Total Cases		Malformed Cases		p Value
	N	%	N	%	
Male	2753	57.5%	68	68%	<0.05
Female	2033	42.5%	32	32%	
Total	4786	100%	100	100%	

Birth Weight of Neonates

The incidence of congenital malformations in low birth weight was 27 (27%) cases of which extremely low birth weight neonates constituted 2(2%), very low birth weight were 9 (9%) and low birth weight were 16 (27.5%). There was association of low birth weight with increased risk of congenital malformations as per Chi-Square test ($p < 0.05$).

Table 3: Birth Weight of Neonates

Birth Weight (kgs)	Total Cases		Malformed Cases		p Value
	N	%	N	%	
<1 kg	14	0.3%	2	2%	<0.05
1-1.5 kgs	95	1.9%	9	9%	
1.501-2 kgs	350	7.3%	16	16%	
2.001-2.5 kgs	1435	29.9%	22	22%	
>2.5 kgs	2892	60.6%	51	51%	
Total	4786	100%	100	100%	

Distribution of cases according to Gestation: The incidence of congenital malformations was significantly higher in preterm neonates as compared to full term and post term neonates ($p < 0.05$).

Table 4: Distribution of cases according to Gestation

Gestation	Total Cases		Malformed Cases		p Value
	N	%	N	%	
Preterm	196	4.1%	12	12%	<0.05
Term	4428	92.5%	86	86%	
Postterm	162	3.4%	2	2%	
Total	4786	100%	100	100%	

Distribution of cases according to Maternal Age

Mothers between 18-20 years had 2% neonates with congenital malformations, while mothers in the age range of 21-25, 26-30 and >30 years had 72%, 18% and 8% neonates with congenital malformations respectively. There was significant association of maternal age and congenital malformations as per Chi-Square test ($p < 0.05$).

Table 5: Distribution of cases according to Maternal Age

Age (yrs)	Total Cases		Malformed Cases		p Value
	N	%	N	%	
18-20	282	5.9%	2	2%	<0.05
21-25	3110	64.9%	72	72%	
26-30	1184	24.8%	18	18%	
>30	210	4.4%	8	8%	
Total	4786	100%	100	100%	

Distribution of cases according to Parity: It was observed that there was significantly higher incidence of

malformation among mothers of gravida 4 or more as per Chi-Square test ($p < 0.05$).

Table 6: Distribution of cases according to Parity

Parity	Total Cases		Malformed Cases		p Value
	N	%	N	%	
Primigravida	1690	35.3%	35	35%	<0.05

2 nd Gravida	1145	23.9%	23	23%
3 rd Gravida	1836	38.4%	34	34%
>4 th Gravida	115	2.4%	8	8%
Total	4786	100%	100	100%

Distribution of cases according to System

The congenital malformations involving Cardiovascular were most common (28%) followed by Renal (14%), Central Nervous system (11%), Musculoskeletal (10%), Genitalia (6%), CTEV (6%), Cleft Lip & Palate (5%),

Dysmorphism (4%), Tracheoesophageal fistula (4%), Ambiguous Genitalia (3%), Single Umbilical Artery (2%), Congenital Diaphragmatic Hernia (2%), Congenital Bowel Obstruction (2%) and Imperforate Anus (2%).

Table 7: Distribution of cases according to System

System	N	%
Cardiovascular	28	28%
Renal	14	14%
Central Nervous System	11	11%
Musculoskeletal	10	10%
Genitals	6	6%
CTEV	6	6%
Cleft Lip & Palate	5	5%
Dysmorphism	4	4%
Tracheo-esophageal fistula	4	4%
Ambiguous Genitalia	3	3%
Single Umbilical Artery	3	3%
Congenital Diaphragmatic Hernia	2	2%
Congenital Bowel Obstruction	2	2%
Imperforate Anus	2	2%
Total	100	100%

CTEV - Congenital Talipes Equinovarus

Distribution and incidence of congenital malformations – Cardiovascular (n=28)

In our study, there were 28 cases that had Cardiovascular congenital malformations - Atrial Septal Defect (17 cases), Patent Ductus Arteriosus (7 cases), Hypoplastic Left Heart Syndrome (2 cases), Transposition of the Great Arteries (1 case) and Pulmonary Stenosis (1 case).

Distribution and incidence of congenital malformations – Renal (n=14)

There were 14 cases that had Renal congenital malformations - Dilated renal pelvis (5 cases), Hydronephrosis (4 cases), Ectopic kidney (2 cases), UPJ obstruction (1 case), Agenesis of one kidney (1 case) and Multicystic kidney (1 case).

Distribution and incidence of congenital malformations – Central Nervous System (CNS) (n=11)

There were 11 cases that had Central Nervous System (CNS) congenital malformations - Hydrocephalus (7 cases), Corpus callosum agenesis (2 cases), Dandy walker (1 case) and Meningocele (1 case).

Distribution and incidence of congenital malformations – Musculoskeletal (n=10)

There were 10 cases that had Musculoskeletal congenital malformations - Syndactyly (5 cases), Polydactyly (4 cases) and Hypoplastic digits (1 case).

Distribution and incidence of congenital malformations – Genitals (n=6)

There were 6 cases that had Genitals congenital malformations- Hypospadias (4 cases) and Congenital hydrocele (2 cases).

Distribution and incidence of congenital malformations– Dysmorphism (n=4)

There were 4 cases that had Dysmorphism congenital malformations - Down syndrome (2 cases), Patau syndrome (1 case) and Crouzon syndrome (1 case).

Distribution of cases according to Outcome

77 (77%) neonates were discharged from hospital while 14 (14%) and 9 (9%) neonates expired or were operated upon respectively.

Discussion

A prospective cross sectional study in 4786 live births was conducted to study the range and occurrence of clinically manifested congenital malformations among live born babies. It was noted that out of the total 4786 live births, the number of babies with congenital malformations was 100.

In the current study, there was 2753 (57.5%) and 2033 (42.5%) male and female births. It was noted that there was considerably higher incidence for congenital malformations was seen in male borns in comparison to female births (68 vs. 32 cases respectively) as per Chi-Square test. This can be correlated to the studies of Vyas R *et al.* [10], Singh A *et al.* [11], Taksande A *et al.* [9], Gandhi MK *et al.* [12] and SarkarS *et al.* [13].

Vyas R *et al.* [10] observational prospective study to assess the incidence of congenital anomalies and its distribution according to various systems found 167 neonates born with anomalies, 82 were still births and 85 were live births. 150 neonates had single system defects and remaining 17 were found to be having multiple system anomalies. Out of total 13,614 births, 431 were still born and out of these 82 were found to have birth defects with the incidence rate of 18.3%. This is much higher than 85 live born neonates having birth

defects out of a total of 13,183 live births -incidence of 0.64%.

In our study, the frequency of congenital anomalies for babies having birth low than normal was 27 (27%) cases, while 2(2%) cases were extremely low birth weight, 9(9%) cases were very low birth weight and low birth weight were 16 (27.5%). There was association of low birth weight with increased threat of congenital malformations as per Chi-Square test which can be found similar to the studies of Gandhi MK *et al.* [6] and SarkarS *et al.* [7].

Gandhi MK *et al.* [12] prospective study on incidence of clinically detectable congenital malformations in new-borns and their association with fetal factors reported out of 5518 new-borns 1227 had birth weight less than 1500 grams and out of them 12 (0.97%) babies were congenitally malformed. Congenital malformations and low birth weight was found to have significant association.

The incidence of congenital anomalies in our study was appreciably higher babies who were born prematurely as compared to full term and post term neonates. This is similar with the study of Gandhi MK *et al.* [12].

It was also noted in the present study conducted that the mothers between 18-20 years had 2% neonates with congenital malformations, while mothers in the age range of 21-25, 26-30 and >30 years had 72%, 18% and 8% neonates with congenital malformations respectively. There was significant association of maternal age and congenital malformations as per Chi-Square test. There was significantly higher incidence of malformation among mothers of gravida 4 or more as per Chi-Square test. This is concordant to the studies of Taksande A *et al.* [8, 2] and SarkarS *et al.* [13].

It was noted in our current study that the congenital malformations involving Cardiovascular were most common (28%) followed by Renal (14%), Central Nervous system (11%), Musculoskeletal (10%), Genitalia (6%), CTEV (6%), Cleft Lip & Palate (5%), Dysmorphism (4%), Tracheoesophageal fistula (4%), Ambiguous Genitila (3%), Single Umbilical Artery (2%), Congenital Diaphragmatic Hernia (2%), Congenital Bowel Obstruction (2%) and Imperforate Anus (2%). These findings were consistent with the studies of Vyas R *et al.* [10], Singh A *et al.* [11], SarkarS *et al.* [13], Kumar V *et al.* [14], Agarwal S *et al.* [15], Roy CA *et al.* [16], Ronya R *et al.* [17] and Copp AJ *et al.* [18].

In our study, there were 28 cases that had Cardiovascular congenital malformations - Atrial Septal Defect (17 cases), Patent Ductus Arteriosus (7 cases), Hypoplastic Left Heart Syndrome (2 cases), Transposition of the Great Arteries (1 case) and Pulmonary Stenosis (1 case). Vyas R *et al.* [10] and Taksande A *et al.* [9] noted similar observations in their studies.

Vyas R *et al.* observational prospective study reported 3 cases of diaphragmatic hernia and 2 cases of congenital heart disease. Only 1 case of diaphragmatic hernia was diagnose [10] and before birth.

In the present study, there were 14 cases that had Renal congenital malformations - Dilated renal pelvis (5 cases), Hydronephrosis (4 cases), Ectopic kidney (2 cases), UPJ obstruction (1 case), Agenesis of one kidney (1 case) and Multicystic kidney (1 case). There were 11 cases that had Central Nervous System (CNS) congenital malformations - Hydrocephalus (7 cases), Corpus callosum agenesis (2 cases), Dandy walker (1 case) and Meningocele (1 case).

There were 10 cases that had Musculoskeletal congenital malformations - Syndactyly (5 cases), Polydactyly (4 cases) and Hypoplastic digits (1 case). This is in consistency with the study of Vyas R *et al.* [10].

In the present study, there were 6 cases that had Genitals congenital malformations - Hypospadias (4 cases) and Congenital hydrocele (2 cases). Similar observations were noted in the study of Vyas R *et al.* [10].

Vyas R *et al.* [10] observational prospective study to assess the incidence of birth defects and their system wise distribution reported 22 cases who had anomalies of GI tract (13.2%) - cleft lip and palate - 16, Gastroschisis-2, omphalocele - 4.8 cases (4.85%) of urogenital anomalies, consisting of ambiguous genitalia (2), bilateral hydronephrosis (1), hypospadias (2), polycystic kidney disease (2) and posterior urethral valve (1). 1 case of hydronephrosis, while two of the cases for ambiguous genitalia, two of the cases of hypospadias, 1 case of polycystic kidney disease and 1 case of posterior urethral valve could not be detected by USG and were diagnosed at birth.

In our study, 77 (77%) neonates were discharged from hospital while 14 (14%) neonates expired and 9 (9%) neonates were operated upon respectively. Which can be seen in the studies done by Vyas R *et al.* [10] and Singh A *et al.* [11].

Vyas R *et al.* [10] observational study reported out of the 70 remaining, 35 (50%) babies were discharged from hospital as stable; 24 babies expired while 6 were operated upon and 5 were transferred to higher centers.

Conclusion

Congenital anomalies in newborns were significantly associated with fetal factors like still birth, prematurity and low birth weight. More emphasis should be given on prevention by regular antenatal care and avoidance of known teratogens and probable teratogenic agents. Antenatal diagnosis, genetic counselling, better diagnostic and management facilities should be provided to improve the outcome.

Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

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