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Study Of Clinically Manifested Congenital Malformations In Live Births

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ABSTRACT

Congenital malformations cause a significant share of infant mortality and morbidity and health care expenditure. This study focusses on the spectrum of clinically manifested congenital malformations among live births at Tertiary care hospital and the incidence of clinically manifested congenital malformations among live births at Tertiary care hospital. A cross-sectional study in 4786 live births is a prospective study of surviving infants born scope and clinical manifestations of congenital malformations. It was noted that the abnormal impact significantly detects a total of 4786 live births, the number of infants with a congenital malformation of 100 stillbirths, infant mortality and child morbidity. The number of congenital infant disabilities is due to the advanced diagnostic technology, especially the increase USG prenatal and neonatal period. In the current study, 2753 (57.5%) and 2033 (42.5%), male and female fertility. Congenital male fertility in the baby was observed to be relatively high (68) as compared to comparative female (32) cases, as per the chi-square test ($p<0.05$). For the prevalence of 27 babies (27 per cent), in our study, congenital abnormalities, though two instances (2 per cent) of low birth weights, 9 (9 per cent) of the children were very low birth weights and 16 (27.5 per cent), of the normal birth weight.

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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 (Sadler, 2000). Congenital malformations have been known and recognised for centuries. It is a problem for research because of the 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 mortality, congenital anomalies are responsible (van Gelder *et al.*, 2010; Lee, 2001).

Consequently, it is essential to have basic epidemiological information of these anomalies. Congenital anomaly rates have also been used for primary health services planning. Previous studies also have shown some connection between congenital heart disease (CHD) and other congenital anomalies and genetic syndromes (Czeizel, 2004).

Depending on the severity of Congenital heart disease diagnosis, the clinical course may range from spontaneous closure of defect as seen with small septal defects to a lifelong need for medical and surgical interventions (Khoshnood *et al.*, 2005; Taksande *et al.*, 2010; Singh and Sinha, 2016). The presence of non-cardiac congenital anomalies also significantly impacts the natural history and clinical

course of CHD as these patients may require medical and/or surgical interventions independent of their cardiac pathology. Epidemiology of CHD is well-described (Abubakar *et al.*, 2015).

The most traumatic experience for a gravid woman, her spouse and the family members is, undoubtedly, the unexpected birth of a deformed child, precipitating feeling of horror, inadequacy and failure in parents.

Research Objectives

The research aims to study the spectrum of clinically manifested congenital malformations among live births at Tertiary care hospital and to analyse the incidence of clinically manifested congenital malformations among live births at Tertiary care hospital.

The outcome of the analysis is early detection for the clinically manifested congenital malformations and thus helping in preventing similar malformations in further pregnancies.

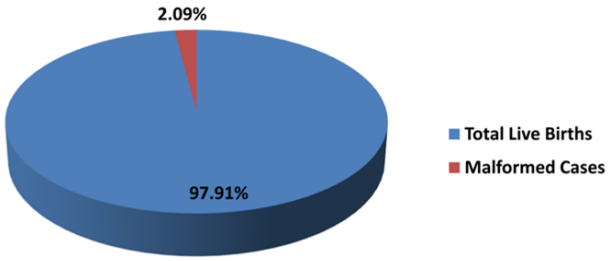

Figure 1: Percentage of malformed cases in Live Births

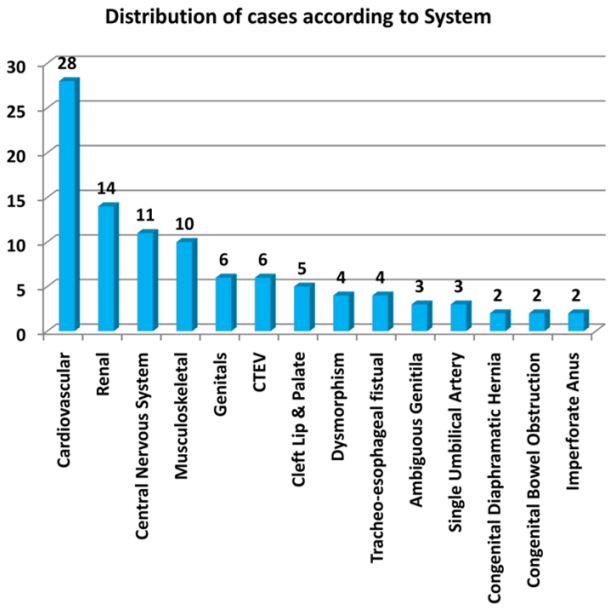

Figure 2: Distribution of cases according to System

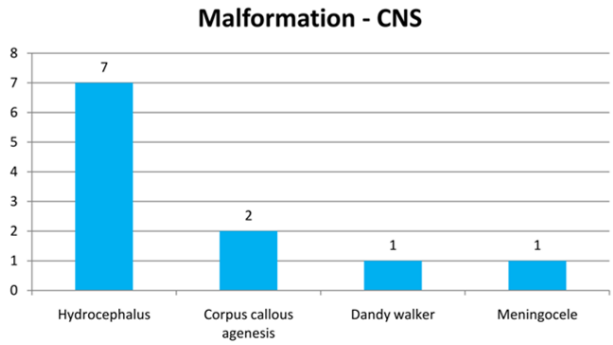

Figure 3: Distribution and incidence of congenital malformations – CNS (n=11)

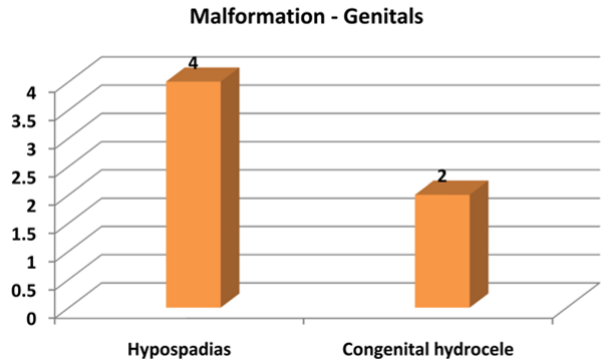

Figure 4: Distribution and incidence of congenital malformations–Genitals (n=6)

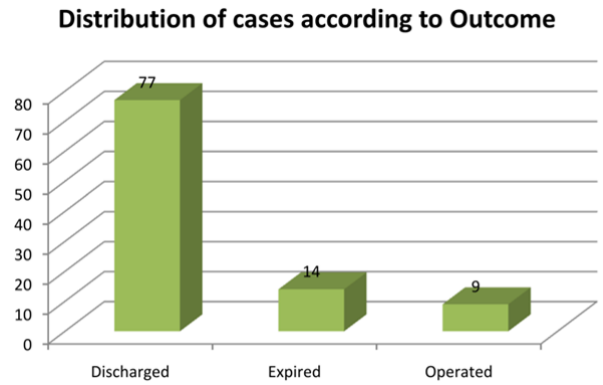

Figure 5: Distribution of cases according to Outcomes

Literature Review

Congenital disorder

A condition existing at the time of birth or before the birth, during the intrauterine period, regardless of the cause is a congenital disorder. From these, those which are characterised by visible or detected on the antenatal scan are termed as —congenital anomalies‖—the WHO defines, Congenital anomalies as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur

Table 1: Gender of Neonates

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

Table 2: Birth Weights of Neonates

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

Table 3: Distribution of cases according to Gestation

Gestation	Total Cases		Malformed Cases		p Value
	N	%	N	%	
Pre-term	196	4.1%	12	12%	<0.05
Term	4428	92.5%	86	86%	
Post-term	162	3.4%	2	2%	
Total	4786	100%	100	100%	

Table 4: Distribution of cases according to Parity

Parity	Total Cases		Malformed Cases		p Value
	N	%	N	%	
Primigravida	1690	35.3%	35	35%	<0.05
2nd Gravida	1145	23.9%	23	23%	
3rd Gravida	1836	38.4%	34	34%	
>4th Gravida	115	2.4%	8	8%	
Total	4786	100%	100	100%	

during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy. In simple terms, congenital refers to the existence at or before birth. So, any substance or factor or insult that causes birth defects are known as teratogens. The complex interactions of the involvement of prenatal insult and post-natal environment play a significant importance in the outcome (Lee, 2001; Czeizel, 2004). Co- relating with the animal research done, studies also suggests that alcohol use, substance abuse, smoking and chemical teratogens also influence the birth outcomes (Singh and Sinha, 2016). Birth defects are present in around 3% of newborns in the USA (van Gelder *et al.*, 2010). Congenital anomalies resulted in about 632,000 deaths per year in 2013 down from 751,000 in 1990 (Ronya *et al.*, 2002).

Epidemiology

Whether the newborn is male or female plays an essential role in the distribution of congenital malformations (Goldman and Schafer, 2016; Patra *et al.*, 2013; Gandhi *et al.*, 2016). There is a male predominance in congenital hypertrophic stenosis. Whereas, it is five times more common for a congenital hip dislocation to occur in female than in the male. A 2010 study established the critical mechanisms of teratogensis (Taksande *et al.*, 2010).

Table 5: 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 fistual	4	4%
Ambiguous Genitalia	3	3%
Single Umbilical Artery	3	3%
Congenital Diaphragmatica Hernia	2	2%
Congenital Bowel Obstruction	2	2%
Imperforate Anus	2	2%
Total	100	100%

*CTEV -Congenital Talipes Equinovarus

Table 6: Distribution and incidence of congenital malformations – Renal (n=14)

Malformation-Renal	N	%
Dilated-renal pelvis	5	36.1%
Hydronephrosis	4	28.4%
Ectopic kidney	2	14.2%
*UPJ obstruction	1	7.1%
Agenesis of one kidney	1	7.1%
Multicystic kidney	1	7.1%
Total	14	100%

*UPJ -Ureteropelvic junction

Table 7: Distribution and incidence of congenital malformations – CNS (n=11)

Malformation-CNS	N	%
Hydrocephalus	7	63.6%
Corpus callous agenesis	2	18.2%
Dandy walker	1	9.1%
Meningocele	1	9.1%
Total	11	100%

Table 8: Distribution and incidence of congenital malformations–Genitals (n=6)

Malformation-Genitals	N	%
Hypospadias	4	66.7%
Congenital hydrocele	2	33.3%
Total	6	100%

Table 9: Distribution of cases according to Outcomes

Outcome	N	%
Discharged	77	77%
Expired	14	14%
Operated	9	9%
Total	100	100%

Oxidant tension, vascular disturbances, folate antagonisms, and basic teratogenesis, which is regulated by particular receptors, or enzymes, which induce about 10% of all congenital defects attributable to adverse exposure to the substance (Vyas *et al.*, 2016). Those involve medications, multiple respiratory diseases, and other workplace contamination that can be avoided. And it was also shown that at least one of the trigger variables was revealed to pregnant people. Flouride when in higher concentrations in water has shown to alter the brain development, causing a significantly low IQ score by around (Singh and Sinha, 2016) points. Carbon monoxide exposure in the developmental stage is responsible for causing skeletal malformations, congenital hip dysplasia, cleft palate and various cardiac defects in the newborn (Zhang *et al.*, 2017). Mothers living near the landfill sites shows a negative impact on the newborn with low birth weight, congenital disabilities, spontaneous abortion, and fetal and infant mortality, which is demonstrated in a study conducted in the United Kingdom. Another issue is the lead exposure, from the majority of the wall paints and pipes containing lead, which is inhaled by the pregnant women, causing learning problems and slow growth and developmental delay (Abubakar *et al.*, 2015).

Patra *et al.* (2013) in 2013 in a cross-sectional descriptive study determined the percentage and types of congenital malformations in live newborns with maternal and perinatal risk factors. The authors found 12,896 babies were born with the prevalence of 2.22% as to 286 were having congenital malformations. 55.7% of women belonged to age between 21 to 30 years. Multiparas had more number of congenital malformations with (3.3%) while primiparas had only 1.8%. Musculoskeletal System (33.2%) being the most common System involved, followed by the gastrointestinal (GI) system (15%). CTEV(17.1%) being the most common one in the musculoskeletal group and likewise cleft lip and cleft palate in the GI system. Congenital anomalies had more association with low birth weight, premature birth, multiple parity and consanguinity.

In a retrospective prenatal review in 2016, Gandhi et al. examined the frequency and relationship of congenital malformations that are clinically observable among adjacent children in a tertiary hospital. Five thousand five hundred eighteen children were born from the authors noticed, 75 of them were identical. The overall number of malformed infants was 68, and the cumulative rate was 1.23 per cent. Two hundred and thirty-six men were unborn, of whom 40 (1.34%) and 28 (1.09%) were congenital malformed out of 2555 people (1.09%). This doesn't impact congenital malformations when the infant is men or women. The impact of these congenital disabilities and their wisely hierarchical structures was examined in 2016 by Vyas *et al.* (2016) in a prospective empirical analysis. Also, 13,614 live / stillbirths were reported by the authors; 167 neonates had congenital malformations. The cumulative frequency was 1.23% (12.3/1000 births). Other structures precede CNS with the most frequent anomaly (53.3%). The investigators established that CNS, including palate, among others, was the most prominent source of these defects.

Zhang *et al.* (2017) in 2017 in a population-based observational study evaluated the time trends in the prevalence of neural tube defects and all their subtypes as well as to identify the epidemiological characteristics of these malformations. The authors found during the observational period, the prevalence of neural tube defects, anencephaly, spina bifida, encephalocele, and congenital hydrocephalus was 19.1, 4.9, 6.2, 1.2, and 9.3 per 10,000 live births, respectively.

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 needed.

Outcome Measures

1. To detect congenital malformations at the earliest and treat correctable causes.
2. To take precautions in the next pregnancy against preventable causes.

Statistical analysis

The appropriate statistical test will be applied as per the requirement of the study. For continuous data, paired 'T-test' will be used to assess the changes. Categorised data will be analysed by the chi-square test. The Fisher's exact test was used when we wanted to conduct a Chi-square test, but one or more of cells had an expected frequency of five or less. Results were graphically represented where deemed necessary. Appropriate statistical software, including but not restricted to MS-Excel. SPSS version 22 was used for statistical analysis. Graphical representation was done in MS-Excel 2016.

Laboratory Investigations

1. Echo-cardiography
2. MRI
3. X-Ray
4. CT Scans
5. Karyotyping

Study Duration

During the study period from October 2016 to March 2018, all live births delivered at Krishna hospital will be examined clinically and investigated further, if required.

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, Figure 1).

Gender of Neonates

There was 2753 (57.5%) and 2033 (42.5%) male and female births (Table 1). Male borns had a higher incidence of congenital malformation compared to females (68 vs 32 cases respectively) as per Chi-Square test ($p<0.05$).

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 an association of low birth weight with increased risk of congenital malformations as per the Chi-Square test ($p<0.05$). Table 2 shows the distribution of birth weights of neonates along with malformed cases.

Distribution of cases according to Gestation

Table 3, the incidence of congenital malformations was significantly higher in pre-term neonates as compared to full-term and post-term neonates ($p<0.05$).

Distribution of cases according to parity

Table 4 it was observed that there was a significantly higher incidence of malformation among mothers of gravida four or more as per the Chi-Square test ($p<0.05$).

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%) in Table 5.

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

Fourteen cases 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 claim) in Table 6 and Figure 2.

Congenital malformations based on – Central Nervous System (CNS) (n=11)

Eleven cases had Central Nervous System (CNS) congenital malformations - Hydrocephalus (7 cases), Corpus callous agenesis (2 cases), Dandy walker (1 case) and Meningocele (1 case) in Table 7 and Figure 3.

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

Six cases had Genitals congenital malformations-Hypospadias (4 cases) and Congenital hydrocele (2

cases) in Table 8 and Figure 4.

Distribution of cases according to Outcomes

Out of total sample population 77 (77%), neonates were discharged from hospital while 14 (14%) and 9 (9%) neonates expired or were operated upon respectively in Table 9 and Figure 5.

CONCLUSIONS

Congenital anomalies in newborns were significantly associated with fetal factors like stillbirth, prematurity and low birth weight. According to the present study, it was observed that 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%). The further focus must also be placed on preventing and avoiding established teratogens and possible teratogenic agents through routine antenatal therapy. To enhance results, antenatal testing, genetic counselling, improved treatment and management should be given. Of avoidance, early detection and also for scheduled termination, routine antenatal visits including prenatal testing, are prescribed if appropriate.

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Nil

Conflict of Interest

I here by declare that there is no conflict of interest related to this manuscript.

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