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Prevalence of Congenital Anomalies in Neonates and Associated Risk Factors in a Tertiary Care Hospital

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Abstract: ***Aims and Objectives: The aim of this study is to determine the proportion and types of congenital anomalies in live newborns and to study associated maternal and perinatal risk factors. Materials and Methods: This cross-sectional descriptive study was carried out in the labour ward and neonatal care unit of a tertiary care center at Civil Hospital, Ahmedabad during the period of April, 2016 to March, 2017. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic variables. Results: During the study period, 7434 live babies were born, of which 166 had congenital malformations, making the prevalence 2.23 %. Most of the women (54 %) belonged to the age group between 21 and 30 years. Congenital anomalies were seen more commonly (3.2%) in the multiparas in comparison with primiparas (1.4%). The predominant system involved was cardiovascular system(28%) followed by CNS (18%), gastro-intestinal (GI) system (17%), musculoskeletal system (15%), genitourinary system (13%), skin (4%), respiratory system (3%) respectively. Congenital anomalies were more likely to be associated with low birth weight (3.2%), prematurity (4.4%), male gender (2.9%), multiparity (3.2%), consanguinity (42%),high maternal age, antenatal history of teratogenic drug intake; radiation exposure; history of certain maternal infections like rubella, TORCH complex and maternal illnesses like diabetes, epilepsy, thyroid disorders during pregnancy and previous history of malformations. Conclusions: Public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended.***

Keywords: Congenital anomalies, low birth weight, prematurity, multiparity, consanguinity

1. Introduction

According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth. However, as per the more recent WHO fact-sheet of October 2012**, congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders), that occur during intrauterine life and can be identified prenatally; at birth or later in life.** Congenital anomalies are an important cause of neonatal mortality both in developed and developing countries. It is not only a leading cause of fetal loss, but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families. With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India. It accounts for **8-15%** of perinatal deaths and **13-16%** of neonatal deaths in India. The pattern and prevalence of congenital anomalies may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables. This study was an attempt to find out the most common system affected and to find out the causal relationship of different etiological factors.

2. Materials and Methods

This cross-sectional descriptive study was carried out in the labour ward and neonatal care unit of a tertiary care center at Civil Hospital, Ahmedabadduring the period of April, 2016 to March, 2017.

Inclusion criteria: All the babies born with congenital anomalies at Civil Hospital, Ahmedabad during this period were included.

Exclusion criteria: Abortions and stillborn were excluded from this study.

The newborns were examined and assessed systematically for the presence of congenital anomalies. Diagnosis of congenital anomalies was based on antenatal ultrasonography-anomaly scan; clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc., System wise distribution of the anomalies was performed.

For each case, a detailed antenatal maternal history including the age of the mother;parity; health and nutritional status of mothers before and during pregnancy; past obstetric history; h/o maternal illnesses like diabetes, epilepsy, thyroid disorders, h/o maternalinfections like rubella, TORCH complex; h/o exposure to radiation and teratogenic agents, h/o consanguinity were obtained by reviewing the

maternal and labour ward records and by interviewing the parents. Socio-economic and family history were reviewed.

A marriage has been considered **consanguineous**, when that is found to have occurred between a male and a female who are blood-related, e.g., between brother and sister, between 1st cousins etc., Birth weights >2.5 kg were considered to be normal; whereas, birth weights <2.5 kg and <1.5 kg were termed as low birth weight (**LBW**) and very low birth weight (**VLBW**) respectively. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as **premature**.

Data was entered into excel data sheet and appropriate statistical analysis was performed. Proportion was calculated and the association was tested with Chi-square test. ***P* < 0.05** was considered to be statistically significant.

3. Results and Discussion

During the study period, **7434** newborns were born in our institution; of which **166** had congenital malformations, making the prevalence **2.23%**. Most common system involved was cardiovascular system (**28%**), followed by CNS(**18%**), followed by gastro-intestinal tract (GIT) (**17%**), musculoskeletal system (**15%**), genitourinary (**13%**) and skin (**4%**). [Table 1.1]

Table 1.1 System wise distribution of congenital anomalies (*n*=166)

System	Number	Percentage (%)
Cardiovascular System	**46**	**28**
Acyanotic heart disease	30	18
Cyanotic heart disease	15	9
Others	1	1
Central Nervous System	**29**	**18**
Meningomyelocele	16	10
Hydrocephalus	6	3.8
Anencephaly	3	1.8
Encephalocele	2	1.2
Others	2	1.2
Gastro-Intestinal System	**28**	**17**
Cleft lip	12	7.2
Cleft palate	5	3.2
Gastroschisis	3	1.8
Tracheo-esophageal fistula (TEF)	2	1.2
Omphalocele	2	1.2
Imperforate anus	1	0.6
Duodenal atresia	1	0.6
Malformation of gut	1	0.6
Others	1	0.6
Musculo-skeletal System	**25**	**15**
CTEV	10	6
Calcaneo-valgus	7	4
Polydactyly	5	3
Syndactyly	2	1.2
Others	1	0.8
Genitourinary System	**21**	**13**
Hydronephrosis	8	5
Ambiguious genitelia	4	2.6
Posterior urethral valve	3	1.9
Polycystic kidney	3	1.9
Hypospadias	2	1
Epispadias	1	0.6
Skin	**6**	**4**
Hemangioma	4	2.6
Skin tag	1	0.7
Aplasia cutis	1	0.7
Respiratory System	**5**	**3**
Diaphragmatic Hernia	4	2.4
Eventration of diaphragm	1	0.6
Sydromes	**4**	**2**
Down	4	2

Talipes (6%) was the most common anomaly seen in the musculoskeletal group and likewise cleft lip (7.2%) and cleft palate (3.2%) in GI system and meningomyeleceole (10%) in CNS. These results are comparable to other national studies. For example-'Prevalence of Congenital Malformations in Indian Maternal Cohort' -first cohort study from India, where 2107 women were followed till pregnancy outcome, in order to measure the prevalence and types of congenital anomalies. Among 1822 births, the total prevalence of major congenital anomalies was 230.51 (170.99–310.11) per 10,000 births. Congenital heart defects were the most commonly reported anomalies in the cohort with a prevalence of 65.86 (37.72–114.77) per 10,000 births.

Table 1.2: Association between Congenital Anomalies and Maternal and Perinatal Risk Factors

Variables	Groups	Congenital Anomalies					
		Yes		No		Total number	X^2 Value, df , P Value
		Number	Percentage (%)	Number	Percentage (%)		
Maternal Age	<20 years	10	0.5	2117	99.5	2127	42.60, df=2, P <0.05
	20-30 years	120	3.2	3894	96.8	4014	
	>30 years	36	2.8	1257	97.2	1293	
Parity	Primiparas	62	1.4	4174	98.6	4236	26.69, df=1, P <0.05
	Multiparas	104	3.2	3094	96.8	3198	
Consanguinity	Present	8	42	12	58	19	114.26, df=1, P <0.05
	Absent	158	2.1	7257	97.9	7415	
Gestation	Term	84	1.5	5492	98.5	5576	53.94, df=1, P <0.05
	Pre-term	82	4.4	1776	95.6	1858	
Mode of Delivery	Vaginal	124	2.6	4708	97.4	4832	7.022, df=1, P <0.05
	Caesarean	42	1.5	2560	98.5	2602	
Gender	Male	119	2.9	3997	97.1	4116	18.235, df=1, P <0.001
	Female	47	1.5	3267	98.5	3314	
Birth Weight	Very low	5	0.5	961	99.5	966	35.71, df=3, P <0.01
	Low	118	3.2	3599	96.8	3717	

	Normal	40	1.7	2264	98.3	2304	
	High	3	0.6	444	99.4	447	

Regarding the parity of the mothers, **4236** were primiparas and rest **3198** were multiparas. Cases of congenital anomaly were found in **3.2%** of multiparas, whereas in primiparas, the proportion was only **1.4%.** It has been seen that more than half of the mothers were aged between 20 and 30 years (**54%**). The prevalence of congenitally anomalous babies born was **0.5%** for mothers <20 years, **3.2%** for 20-30 years and **2.8%** for>30 years. In the present study, **19** mothers had a history of consanguinity and **8** of them showed some congenital anomaly (**42%**) in their babies. Prematurity and Low birth weight (LBW) was found to have a higher risk of congenital anomalies. The occurrence was about **three times more** in case of preterm delivery as compared with the term ones, making it statistically significant.Among all the newborns, **286** babies were born of twin delivery, **3** of triplet delivery and **15** of these **295** babies, which were products of multiple gestations, had one or more congenital anomalies. The congenital anomalies affected significantly higher proportion of male babies (**2.9%)** than their female counterparts (**1.5%**).[Table 1.2].

Advanced diagnostic technology can detect a large number of anomalies in neonatal period. Out of these 166 cases, about 60% of cases were detected after delivery; this was due to poor rate of antenatal screening.

With regards to the associated conditions along with congenital malformations, most common condition associated was abnormalities of amniotic fluid volume.

Despite the high risk of recurrence of congenital malformations, there are no well-accepted preventive measures in developing countries like India. It indicates that strong preventive measures for congenital anomalies in this region are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

4. Conclusion

This study has highlighted the prevalence and types of congenital anomalies seen in our locality.Inadequacies in periconceptional maternal nutrition – folic acid and iodine; are associated with the high incidence of CM. Community members should also be sensitized against early pregnancy and educated to minimize the risk.Congenital malformation, one of the important causes of infant mortality and morbidity can be reduced by proper preconception and antenatal care and second trimester anomaly scan or level two scan. Even the treatment and rehabilitation of these anomalous children is a challenging task. Parents are likely to be anxious and guilt on learning the existence of a congenital anomaly and require sensitive counseling.So,Regular antenatal visits and prenatal diagnosis by early second trimester screening test are recommended for prevention, early intervention and even planned termination, when needed.

5. Limitations

As Civil Hospital, Ahmedabad is a tertiary care hospital or referral center, prevalence calculated may be higher than the general population.Tertiary care hospital usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, the data cannot be projected to the general population, for which population-based studies are necessary. Secondly, we could not include the abortions and stillborn, because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted and in most of the cases, parental consent is not available for pathological autopsy.

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