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Prevalence of Congenital Malformations: A Hospital-Based Study

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Abstract: ***Background: Birth defects are a diverse group of disorders with prenatal origin that can be caused due to single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. The objective of this study was to study the prevalence of congenital anomalies in Department of Pediatrics at Government Regional Hospital Kullu (R.H.Kullu) Himachal Pradesh. Methods: The study population includes all neonates (inborn plus outborn) in R.H. Kullu in one year i.e from 1st August 2017 to 31st July 2018. The babies were examined and assessed thoroughly for the presence of a congenital anomaly and were then distributed system wise. Results: Among the 3317 deliveries, 92 babies had congenital malformations. Consanguinity and increased maternal age were found to increase the presence of congenital anomalies. Cardiovascular and urogenital malformations were found to be the most common. Conclusions: Congenital anomalies are a global health problem. This study supports us to find out the caause of congenital anomalies. Consanguinity should be discouraged. Early antenatal scan aids in prior detection of congenital anomalies and appropriate genetic counselling can reduce the anomalies in future pregnancies.***

Keywords: Congenital malformations, Prevalence

1. Introduction

A congenital anomaly is any alteration, present at birth, of normal anatomic structure. It may be major or minor, isolated or part of a larger constellation of defects, of clear or uncertain cause. Several genetic and environmental etiologies are well delineated , but the fundamental etiology of nearly half of all birth defects is unknown .[1]

Birth defects, congenital abnormalities and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects that are present at birth[1]. According to WHO Fact sheet, out of all causes of 2.761 million deaths worldwide during the neonatal period in 2013, congenital malformations contributed to 276000 deaths, preterm birth complications 965000, intra partum related complications (birth asphyxia) 662000, neonatal sepsis 421000 and other important causes 437000[2] .

Congenital malformations can be caused by varied causes such as multifactorial inheritance (23%), familial (14.5%), chromosomal disorders(10.1%), single mutant gene (3.1%), environmental teratogens(3.2%) ,uterine factor (2.5%) and twinning (0.4%) and other causes such as maternal infections , systemic illnesses [3]. These birth defects tend to recur at a low rate, approximately 3% to 5% for each subsequent pregnancy for the parents of one affected child, 10% to 15% if two siblings have previously been similarly affected[4].

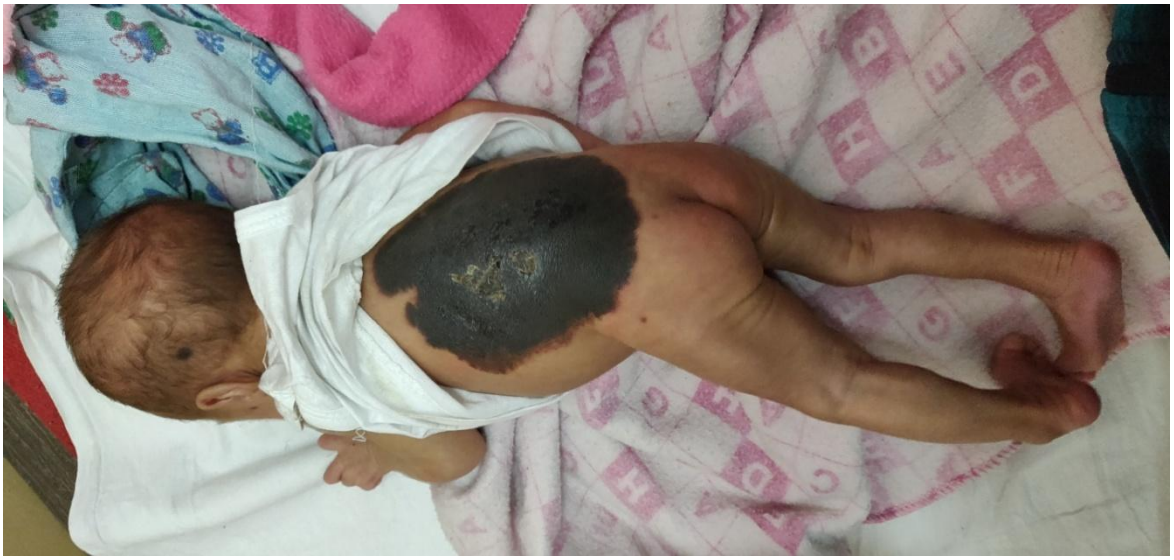
Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India[5,6] .The proportion of perinatal deaths due to congenital malformations is increasing in world as a result of reduction of mortality due to other causes leading to the improvement in perinatal and neonatal care. The present study was carried out with the aim to determine the prevalence of congenital malformations, as well as incidence of affecting various organ systems at our hospital over a period of one year.

2. Methods

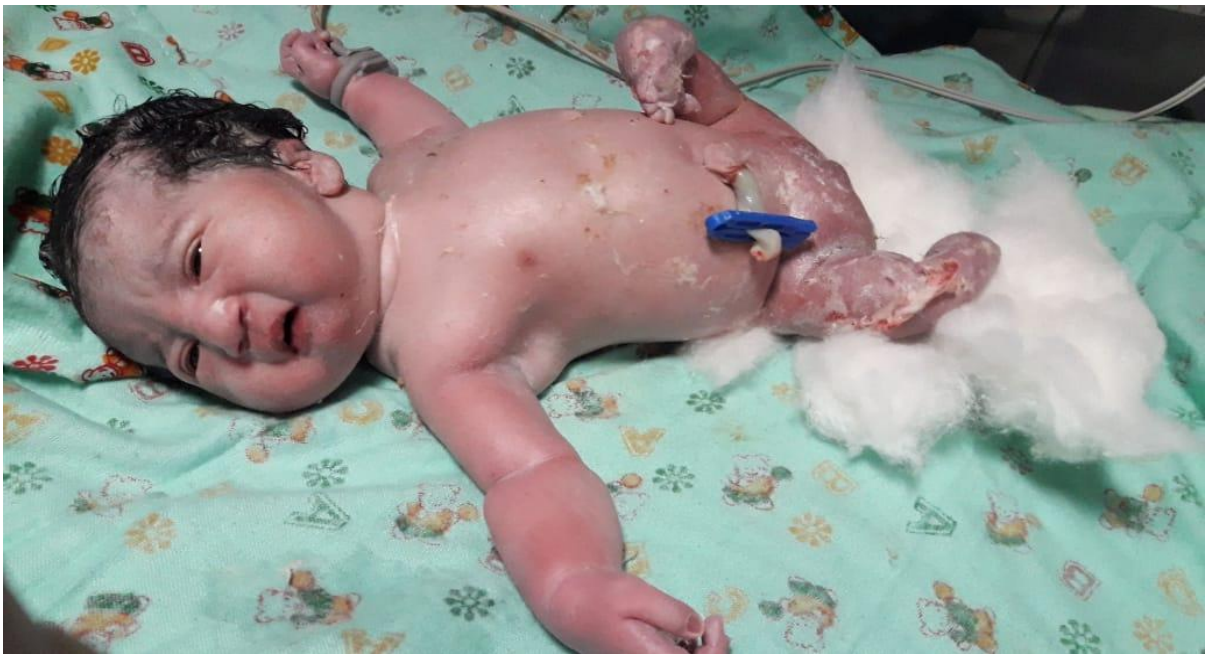
This prospective study was done at Regional Hospital Kullu. All neonates (inborn plus outborn) born from August 2018 to July 2019 were included in the study. Babies were examined by pediatrician at the time of birth and follow up was done till discharge/referral. A detailed history was taken including familial and gestational factors, and meticulous examination of neonates were done. All neonates identified with anomalies were further investigated. Radiographs, ultrasound examinations, neurosonogram, echocardiography, and chromosomal studies were done wherever necessary. The surgical conditions were evaluated and then treated appropriately/referred to PGIMER, Chandigarh. The Institutional ethical committee approval was received.

3. Results

During this 1-year study 3377 newborns were delivered, which included 60 IUD, 40 twin gestations and 92 had one or more congenital anomaly. The prevalence rate of CMF came out to be 2.78%. The pattern of congenital malformations seen in neonates; most commonly affected urogenital (19.56%) and cardiovascular system (17.40%).



Congenital nevus



Syndromic baby

Von hippel lindau syndrome

Meningomyelocoele

Table 1: Demographics

Total no of singleton delivery	3377
Twin delivery	40
Delivery by lscs	450
IUD(still birth/macerated baby)	60
Newborn with CMF	92

Table 2: Gender Distribution of Newborn with Birth Defects

Gender	Number	Percentage
Male	48	52.17
Female	44	47.83

Table 3: Socio-Demography of Mother and Neonates in Study

		All mothers	Mothers with CMF babies	Incidence of CMF (%)
Mothers	18-23 years	1227	27	2.20
	24-29 years	1635	20	1.22
	>30 years	455	45	9.90
Parity	1	609	50	8.21
	2	1428	25	1.75%
	3	740	7	0.94%
	>4	540	10	1.85%
ANC	Booked	3315	92	2.78%
	Unbooked	2	0	0.0%
Residence	Rural	2337	82	35.1%
	urban	980	10	64.9%
Consanguineous Marriage		9	6	66.67
Family History of CMF Baby		4	1	25
Previous Child with CMF		7	1	14.29

As evident from above table, increased maternal age was associated with increased incidence of CAs. This was primarily more in mothers >30 years of age. There was significantly more CAs among neonates with parental consanguinity than among babies without parental consanguinity.

Table 4: Association Between Gestational Age and Congenital Anomalies

Gestational age	Congenital anomaly	Percentage
Preterm	58	63.05
Term	34	36.09

Table 5: System Wise Distribution of Congenital Anomalies:

System	Total No.	Malformation	No.	Percentage
CNS	12	Meningocoele	3	13.04
		encephalocoele	2	
		anencephaly	2	
		hydrocephalous	2	
		microcephaly	3	
CVS	16	Patent ductus arteriousus	8	17.40
		ASD	2	
		VSD	5	
		TOF	1	
Urogenital	18	Ambiguous genitalia	0	19.56
		hydronephrosis	5	
		hypospadiasis	3	
		hydrocoele	10	
GIT	14	Cleft lip/palate	8	15.21
		CHPS	3	
		TEF	1	
		Imperf anus	2	
		CDH	0	
MSK	12	polydactly	9	13.04
		CDH	3	
skin	11	Nevus	2	11.96
		Sacral dimple	9	
miscellanous	9	Down's syndrome/ syndromic baby	4/5	9.79

4. Discussion

The fundamental approach to managing an infant with one or more congenital anomalies is much the same as the management of any other clinical scenario. Effective clinical intervention is organized around an understanding of the natural history of the condition at hand. History taking begins with conception and includes a detailed three-

generation pedigree. Physical features must be scrutinized, measured, and documented with precision, and confirmatory studies must be carefully chosen and accurately interpreted

In our hospital based prospective study, the overall prevalence of congenital malformations was 2.7% (92 of 3317) of live born neonates and the most common system involved were CVS and Genito urinary system.

There are variations in prevalence of congenital malformations in different parts of the world which might be explained by social and racial influences commonly known in genetic disorders. Also, the results may vary according to the background of the investigators, the type of sample studied and the period of observation..The annual report of Indian Medical Research says that the commonest congenital malformation is cardiac in nature (0.57%). [16]

The current study found that congenital malformations commonly prevailed in babies born to consanguinous marriage. History of consanguinity was found to be present in about 66.67% in the present study. The role of parental consanguinity for the development of congenital malformations has been addressed by other studies.[9-12] On the other hand, gender of the babies was not significantly associated with the development of congenital malformations. In Saudi Arabia, Al shehri reported a high frequency of major congenital malformations and stated that it might have resulted from the common habit of consanguineous marriages which has led to the preservation of rare mutations[11].

Our study has statistically shown that mothers, above 30 years of age, are at a higher risk of producing malformed babies. Sugunabai, reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Datta et al, documented statistically insignificant association of increased maternal age and congenital anomalies.[7,15]

The incidence of congenital malformations has no association with LBW in the present study. This association of LBW and malformations has been well documented in other studies.[8-10]

Many studies have documented a male preponderance among congenital malformed babies.[9-12] However, in the present study we could not observe any major difference in predilection of malformations according to gender. On the other hand, Gupta et al, reported that the incidence of congenital musculoskeletal malformations was apparently found to be higher in female babies than in males; however, the difference was not statistically significant.[13]

Regarding the gestational age of the malformed neonates, we found a significantly increased incidence of congenital malformations among preterm neonates than full term. This is in accordance with reports by others.[5,14] Jones added that the risk factors associated with prematurity has proven increased frequency of CMF.

In our study we observed a high incidence of Neural Tube Defects (13.04%) which can be prevented by early prenatal diagnosis .

Besides larger multicentric studies are needed to determine the exact congenital anomaly distribution of our country. Widespread health education in the population and pregnant females can help in preventing many etiological factors of congenital malformations .

5. Conclusion of the Study

Most children who are born with major CMF and survive infancy are affected physically, mentally or socially, or can be at increased risk of morbidity due to various health disorders. Thus primordial and primary prevention are vital to decrease incidence of CMF and the morbidity associated with it.

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Ethical approval: The study was approved by the institutional ethics committee

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