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Original Research Article

Prevalence of congenital anomalies: 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 by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and or micronutrient deficiencies. The objective of this study was to study the prevalence of congenital anomalies in Department of Pediatrics at Rajarajeswari Medical College and Hospital, Kambipura, Bangalore.
Methods: The study population includes all babies born between 1 August 2015 and 31st July 2016. The babies were examined and assessed thoroughly for the presence of a congenital anomaly and were then distributed system wise by the pediatrician. Surgical conditions were also reevaluated by a pediatric surgeon.
Results: Among the 2,137 deliveries, 86 babies had congenital malformations. Prematurity, consanguinity and increased maternal age elevate the appearance of congenital anomalies. The cardiovascular malformations were most common with a prevalence rate of 4%.
Conclusions: Congenital anomalies are a global health problem. Thus, this study supports us to understand the prevalence of congenital anomalies. There is no association of congenital malformations with low birth weight babies. Consanguinity should be discouraged. Early antenatal scan aids in prior detection of congenital anomalies Appropriate genetic counselling can reduce the anomalies in future pregnancies.

Keywords: Congenital malformations, Prevalence

INTRODUCTION

Birth defects are a diverse group of disorders with prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and or micronutrient deficiencies. Maternal infections such as rubella, CMV systemic illnesses like diabetes mellitus (DM), hypothyroidism and folic acid deficiency, exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals and doses of radiation are all other factors that cause birth defects.[2] Birth defects, congenital abnormalities and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects that are present at birth.[3]

Congenital malformation represents defects in morphogenesis during early fetal life. According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.[4] The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[5,6] Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the pediatrician. The proportion of perinatal

deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centers providing good neonatal care. The present study was carried out with the aim to determine the prevalence congenital malformations, as well as incidence affecting various organ systems, at our hospital.

METHODS

This prospective study was undertaken at Rajarajeswari Medical College and Hospital. All babies (inclusive of twins) born from August 2015 to July 2016 were included in the study. Babies were examined by pediatrician at the time of birth and within 24 hours of birth and were further followed up to 72 hours. A detailed history was taken including familial and gestational factors, and a meticulous examination of baby were done. All neonates identified with CAs were further investigated. Radiographs, ultrasound examinations, neurosonogram echocardiography, and chromosomal studies were suggested wherever necessary. The surgical conditions were re-evaluated by pediatric surgeon and then treated appropriately. The results were analyzed by simple statistical techniques such as recording number and percentage of cases. Student's t-test was applied and P <0.05 was considered significant. The Institutional ethical committee approval was received.

Figure 1: Collodion Baby

Figure 2: Multiple anomalous baby.

Figure 3: Anencephaly.

Figure 4: Ambiguous genitalia.

RESULTS

During this 1-year study 2,137 newborns were delivered, which included 40 IUD, 14 twin gestations, (4 had CVS anomalies) and 86 had one or more congenital anomaly. The prevalence rate is 4%. The pattern of congenital malformations seen in neonates; most commonly affected cardiovascular system (41.86%), followed by the central nervous system (18.62%), gastrointestinal system (15.11%), genitourinary system (11.62%), skin (6.97%), musculoskeletal system (5.35%), and syndromic (2.34%).

Table 1: Demographics.

Deliveries and births	Number
Total no. of singleton deliveries	2123
Total no. of twin deliveries	28
Total no. of babies born	2151
Total no. of malformed babies	86

Table 2: Gender distribution of birth defect.

Gender	Number	Percentage (%)
Male	46	53.5%
Female	40	46.5%
Total	86	100%

p >0.05 not significant.

Maternal age was associated with increased incidence of CAs although this was primarily in mother more than 30 years. There was significantly more CAs among neonates with parental consanguinity than among babies without

parental consanguinity (P<0.05). The prevalence of CAs was significantly higher among the preterm babies than among the term babies. However, the gender of the baby plays no role in the frequency of CAs.

Table 3: Association between age of mother and congenital anomaly.

Age of mothers in years	Congenital anomaly	Percentage (%)
18-23 years	26	30.2
24-29 years	10	11.6
Above 30 years	50	58.2
Total	86	100

df = 1, x^2 = 0.6; p <0.05, significant

Table 3 summarizes the maternal and fetal factors associated with congenital anomaly at birth. Maternal age was associated with increased incidence of congenital anomaly. Although this was significant, more congenital anomaly is seen in mother more than 30years.

Table 4: Association between Consanguinity and congenital anomaly.

Consanguinity	Congenital anomaly	Percentage (%)
1st degree	71	82.5
2nd degree	08	9.3
3rd degree	07	8.2
Total	86	100

df = 1, x^2 = 0.5; p <0.05, significant

Table 5: Association between gestational age and congenital anomalies.

Gestational age	Congenital anomaly	Percentage %
Preterm	63	73.2
Term	23	26.8
Total	90	100

df = 1, x^2 = 0.4; p <0.05, significant

Table 5: System wise distribution of congenital anomalies.

System type	Total no	Malformation	No.	Percentage (%)
CVS	36	Patent ductus arteriosus	15	41.86%
		Atrial septal defect	12	
		Ventricular septal defect	06	
		Tetralogy of fallots	03	
CNS	16	Meningomyelocele	04	18.62%
		Encephalocele	03	
		Anencephaly	05	
		Blakes cyst	01	
		Choroid plexus cyst	01	
		Mesencephalic cyst	01	
		Communicating frontenella	01	
Urogenital system	10	Amibigious genitalia	01	11.62%
		Hydronephrosis	02	
		Hypospadius	01	
		Hydrocele	03	
		Undesended testis	03	
GIT	13	Tracheo-esophageal fistula	01	15.11%
		Diaphragmatic hernia	01	
		Cleft palate	04	
		Imperforate anus	03	
		Cleft lip	04	
Musculoskeletal	03	Polydactyly	02	3.48%
		Syndactyly	01	
Skin	06	Collodion baby	01	6.97%
		Pre-auricular tag	03	
		Scaral dimple	02	
Miscellaneous/syndromes	02	Down	01	2.34%
		Others	01	
Total	86			100%

Table 4 shows association between congenital anomalies and consanguineous marriage. First degree consanguineous marriage has significant increase in congenital anomaly than second degree and third degree.

The risk for preterm was significantly higher in pregnancies with multiple congenital anomaly than in term with single anomaly.

DISCUSSION

With improved control of infections and nutritional deficiency diseases, CAs have become important causes of perinatal mortality in developed countries and will very soon become increasingly important determinates of perinatal mortality in developing countries.[8] There is currently no reliable statistical data of the number of babies who were born with a serious congenital disorder due to genetic or environmental causes. In our hospital-based prospective study, the overall prevalence of CAs was 4% (86 of 2137) of live born neonates. There are variations in prevalence of CAs in different parts of the world might be explained by social and racial influences-commonly known in genetic disorders.

Also, the results vary according to the background of the investigators, the type of sample studied and the period of observation. In this study, the most common system involved was the cardiovascular system (41.86%), the central nervous system (18.62%), gastrointestinal system (15.11%), genitourinary system (11.62%), skin (6.97%), musculoskeletal system (5.35%), and syndromic (2.34%). The annual report of Indian Medical Research 16 says that the commonest congenital malformation is cardiac in nature (0.57%).

The current study found that CAs commonly prevailed in babies born to consanguineous marriage. History of consanguinity was about 82% in the present study. The role of parental consanguinity for the development of CAs has been addressed by other studies.[9-12] On the other hand, sex of the babies was not significantly associated with the development of CAs. In Saudi Arabia, Alshehri reported a high frequency of major CAs and stated that it might have resulted from the common habit of consanguineous marriages which has led to the preservation of rare mutations. This 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.[15,7]

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 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 CAs 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 Ca's.

Fetal autopsy was not done, as the patient relatives did not give consent. Most of the internal anomalies could not be identified by visual examination.

CONCLUSION

Congenital anomalies are a global health problem. Thus, this study supports us to understand the prevalence of congenital anomalies. There is no association of congenital malformations with low birth weight babies. Consanguinity should be discouraged. Early antenatal scan aids in prior detection of congenital anomalies. Appropriate genetic counselling can reduce the anomalies in future pregnancies.

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