Research Article

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Spectrum of congenital malformations in newborns: in a medical college hospital in South India

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ABSTRACT

Background: The study was carried out with the aim to determine the overall rate of congenital malformations, incidence in live births and stillbirths, as well as incidence affecting various organ systems, at a medical college hospital in Karnataka and compare them to previous studies

Methods: All intramural deliveries between June 2012 and March 2014 were included in the study. All the new-borns were examined for congenital malformations soon after birth and every day during routine ward rounds. Relevant radiological, histo-hematological and genetic tests were carried out.

Results: During the study period there were 3016 births out of which 2941 were live births and 75 were still born. 93 babies had one or more malformations. The overall incidence of malformations was 3.083%. Incidence of malformations among live births was 2.72% whereas it was 17.33% among still born babies. Musculoskeletal malformations accounted for 27.5% of all the malformations. This was followed by cutaneous 19.16%, genitourinary 15.83%, gastrointestinal 12.5%, neurological 10% and cardiac 5.83%.

Conclusion: Congenital anomalies are a major cause of stillbirths and infant mortality. Musculoskeletal malformations were the commonest system involved. Still born babies had a significantly higher incidence of malformations and also had a higher incidence of multiple malformations.

Keywords: Congenital malformations, New-born, Still born

INTRODUCTION

A congenital anomaly may be narrowly defined in terms of physical structure as a malformation, an abnormality of physical structure or form usually found at birth or during the first few weeks of life;¹ or defined more widely to include functional disturbance as a defect, any irreversible condition existing in a child before birth in which there is sufficient deviation in the usual number, size, shape, location or inherent character of any part, organ, cell or cell constituent to warrant its designation as abnormal.² A congenital anomaly is thus any alteration present at birth of normal anatomic structure and has cosmetic, medical or surgical significance.

According to WHO, the term "congenital malformations" should be confined to structural defects present at birth.⁴ Congenital malformations account for 8-10% of all perinatal deaths and 13-16% of all Neonatal deaths.³ With improvement in perinatal and neonatal care, birth defects will become leading cause of neonatal mortality and morbidity.² As other causes of infant mortality like infections and nutritional deficiencies are being brought under control, congenital malformations are rapidly emerging as one of the major worldwide problem.^{5,6} For more than two decades, congenital anomalies have been the leading cause of infant mortality in the United States.⁹ In spite of the frequency of congenital anomalies the underlying causes for most remain obscure. Around 40%-60% of congenital anomalies are of unknown etiology.^{9,10}

The prevalence rate of congenital anomalies is increasing due to exposure of teratogens of various kinds.¹¹ In India congenital malformations have emerged as the third commonest cause of perinatal mortality.

The present study was carried out with the aim to determine the overall rate of congenital malformations, incidence in live births and stillbirths, as well as incidence affecting various organ systems, at a medical college hospital in Karnataka and compare them to previous studies.

METHODS

This study was conducted in department of pediatrics at Basaveshwara Medical College Hospital, Chitradurga, Karnataka. All the intramural deliveries between June 2012 to March 2014 comprised the study material. There were a total of 2941 live births and 75 stillbirths during this period. All the newborns were examined for congenital malformations soon after birth and every day during routine ward rounds. Relevant information regarding maternal age, gestational age, sex, community, birth weight, birth order and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Antenatal ultrasonography (USG) findings were noted. Relevant radiological, histo-hematological and genetic tests were carried out. Autopsy was done on stillbirth and neonatal death, whenever parents consent could be obtained. Karyotyping was done. A meticulous general and systemic examination was carried out by a consultant at the time of birth to detect any malformations. Ultrasound was employed routinely to detect multiple congenital anomalies and to rule out majority of the internal congenital anomalies. 2D echocardiography was also used for all congenital heart diseases, along with the routine X-ray chest and electrocardiogram. Computed Tomography (CT) scan were advised only for certain special cases. Malformations were divided into major and minor; major malformation interferes considerably with the function of all or part of the infant, minor malformation gives no serious medical or cosmetic consequences to the patients. The major malformations were divided into Central Nervous System (CNS), musculoskeletal, gastrointestinal, genitourinary, Cardio-Vascular System (CVS), syndromes and miscellaneous disorders. Statistical analysis was done using Z test and Chi-square test.

RESULTS

During the study period there were 3016 births out of which 2941 were live births and 75 were still born. 93 babies had one or more malformations. The overall incidence of malformations was 3.083%. Incidence of malformations among live births was 2.72% whereas it was 17.33% among still born babies. Still born babies had a significantly higher incidence of malformations and

also had a higher incidence of multiple malformations (Table 1).

Table 1: Profile of study population.

Details	Numbers	Percentage
Total births	3016	
• Live births	2941	97.51
• Still births	75	2.4
Neonates with malformations	93	3.083
• Live births	80	2.72
Still births	13	17.33
Total malformations	120	3.97
• Live births	106	3.60
• Still births	14	18.66

Musculoskeletal malformations were the commonest malformation and accounted for 27.5% of all the malformations. This was followed by cutaneous 19.16%, genitourinary 15.83%, gastrointestinal 12.5%, neurological 10% and cardiac 5.83% (Table 2).

Table 2: Distribution of congenital malformations.

System	Number	Percentage
Musculoskeletal	33	27.5
Cutaneous	23	19.16
Genitourinary	19	15.83
Gastrointestinal	15	12.5
Neurological	12	10
Cardiac	7	5.83
Spine	4	3.33
Eyes	2	1.66
Others	5	4.16

Musculoskeletal and cutaneous malformations were more common among live births, whereas central nervous system malformations were more common among still births.

DISCUSSION

The incidence of congenital malformations in this study is 3.083%. Singh et al. from India who reported a frequency of 1.5% and Golalipour et al. from Iran (1.01%).^{15,16} Desai et al. from Bombay, India, Fatema et al from Bangladesh found a little higher incidence of 3.61% and 3.68% respectively.^{17,18}

The slightly higher incidence of congenital malformations in our study can be attributed to the low socioeconomic status of the majority of the patients, poor antenatal care. Many of the patients had not undergone antenatal ultrasonography. Other factors include differences in study protocol, geographical and race. Frequency of congenital malformation was slightly higher in males in our series. Congenital anomalies were

seen more in young and older mothers. Similar findings were observed by others.

One study from India and another from Iran found musculoskeletal anomalies as highest in order (30.60% and 30.10% respectively).^{15,16}

Fatema et al. from Bangladesh and Tomatir et al. from Turkey found that central nervous system abnormality were the highest in position in their studies (46.67%, 31% respectively).^{18,22}

The low prevalence of cardiovascular defects at birth is due to the fact that most CHD's become symptomatic by 2-4 months of age. Among congenital heart anomalies VSD was the most common. In chromosomal anomalies Down's syndrome was most frequently seen. Congenital talipes equino varus was the commonest musculocutaneous abnormality observed in our study. Among the genitourinary tract anomalies, hypospadias, undescended testis, and polycystic kidney were the most prevalent lesions.

Consanguinity of marriage, maternal exposure to some drugs, maternal disease, maternal smoking habit have some relation with congenital malformations.

CONCLUSION

Congenital anomalies are a major cause of stillbirths and infant mortality. By thorough clinical examination, the life-threatening congenital malformation must be identified, as early diagnosis and surgical correction of the malformed babies offer the best chance for survival. Stringent antenatal ultrasonography may be able to pick up major, life threatening congenital anomalies.

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