

Research Article**Antenatal and Perinatal Profile of Patients with Birth Defects in a District Hospital****Vijayalakshmi. P¹, Venugopal. S^{2*}, R. B. Patil³**¹Junior Resident, Department of Paediatrics, Shimoga Institute of Medical Science, Shivamogga, Karnataka, India²Assistant Professor, Department of Paediatrics, Shimoga Institute of Medical Science, Shivamogga, Karnataka, India³Professor & HOD, Department of Paediatrics, Shimoga Institute of Medical Science, Shivamogga, Karnataka, India***Corresponding author**

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Abstract: The aim was to study the antenatal and perinatal profile of infants and paediatric patients with birth defects (BDs). The present study was a single center retrospective study. Infants and paediatric patients with birth defects admitted in Neonatal intensive care unit (NICU), Paediatric intensive care unit (PICU) and Paediatric wards from January 2014 to December 2014 were enrolled. The study was performed over a period of 12 months from January 2014 to December 2014. During the study period, total paediatric admissions were 5982 including paediatric intensive care and neonatal intensive care units. Of these 276(4.61%) patients had birth defects. A total of 457 birth defects were identified in these 276 patients. The mean maternal age at conception in the present study was 27.2 years (age range of 16 to 38 years and standard deviation of 4.2 years. Primigravida mothers constituted 33.69% of cases followed by 2nd (26.81%) and 3rd (22.83%) gravida. BDs were detected antenatally in only 38.45% of cases, whereas ultrasound was not done in one fourth of cases. Mothers of 35.87% (n=276) of cases had history of abortions and mothers of 7.25% (n=276) of cases had history of multiple abortions. Forty three cases (15.57%, n=276) had siblings with BD. The rate of dissimilar BD in the siblings (13.04%, n=276) is more compared to similar BD (2.53%, n=276). 151 cases (54.71%) had normal birth weight and 115 cases (41.67%) had low birth weight. 114 cases (41.3%) had multiple birth defects and 162 cases (58.7%) had isolated birth defects. Cases of isolated BDs (n=162) most commonly involved musculoskeletal system (41.36%) followed by cardiovascular system (24.08%) and central nervous system (22.22%). Birth defects were more common in primigravida mothers with the age group of 25 – 30 years. Not all the birth defects were detected antenatally. Mothers of patients with birth defects had significant rates of previous abortions. The rate of having dissimilar BD in the siblings was more than similar birth defect. BD cases more commonly had normal birth weight. Isolated birth defects were more common than multiple BDs but the rate of multiple BDs was significantly high. The musculoskeletal system was the most common system involved.

Keywords: Birth Defects (BDs), Antenatal profile, Perinatal profile.

INTRODUCTION

The toll of birth defects worldwide has been recognized as a severe public health problem. Birth defects, affecting 2-3% of all infants, are a major cause of perinatal mortality and childhood morbidity in both developed and developing countries [1, 2]. The decline in infant and childhood mortality rates in most countries in the 20th century is a public health triumph. Improvements in socioeconomic, educational, and health care conditions, and the strengthening of infrastructure in high-income countries, began in the first half of the last century and led to significant improvements in health [3]. The “health transition” is initially marked by a decline in infant and under-5 mortality from infectious diseases and malnutrition, which predominate in the early years of life [4]. At the same time, however, mortality from birth defects has

remained constant. As a result, birth defects assume a greater proportional cause of infant mortality as countries develop.

The goal of a dysmorphology assessment is to interpret the pattern of structural anomalies correctly and arrive at the diagnosis conclusively. This diagnostic search is motivated primarily by the immediate need to provide the patient and the family with accurate information about the prognosis and natural history of the condition and its response to treatment. Knowledge of the diagnosis also allows the physician to perform a directed search for occult anomalies and therapeutic decisions in consultation with the family. In the longer term, the heritability recurrence risks, and availability of prenatal diagnosis for the condition in question depend on making the correct diagnosis [5].

Aim and objectives

To study the antenatal and perinatal profile of infants and paediatric patients with birth defects (BDs).

MATERIALS AND METHODS

The present study was a single center retrospective study. Infants and paediatric patients with birth defects admitted in Neonatal intensive care (NICU), Paediatric intensive care (PICU) and Paediatric wards from January 2014 to December 2014 were enrolled.

Inclusion criteria

- Age day 1 to 12 years
- Defects occurring and/ or presenting at birth or recognized later in infancy or childhood.
- Only those cases where the birth defects were obvious or their nature conclusively proven by appropriate investigations were included for analysis.

Exclusion criteria

- Age > 12 years.
- Defects secondary to complications and sequels of conditions such as prematurity (patent ductus arteriosus) or infections acquired in the post-natal period (hydrocephalus).
- Patients admitted to other speciality wards.
- Still births and intrauterine deaths.
- Death within 8 hours of admission.

RESULTS

During the study period, total Paediatric admissions were 5982 including Paediatric intensive care and Neonatal intensive care units. Of these, 276(4.61%) patients had birth defects. A total of 457 birth defects were identified in these 276 patients. Details of mothers' antenatal profile and perinatal profile of patients with birth defects are shown in table 1 and table 2 respectively.

Table 1: Distribution of patients with respect to mothers' antenatal profile (n=276)

Parameter	No. of cases (n=276)	Percentage	
Maternal age at conception	18 – 25 years	82	29.71%
	26 – 30 years	110	39.86%
	31 – 35 years	78	28.26%
	>35 years	6	2.17%
Mothers' gravida	Primigravida	93	33.69%
	2 nd gravida	74	26.81%
	3 rd gravida	63	22.83%
	4 th and >4 th gravida	46	16.67%
Antenatal USG	Done and BD detected	106	38.41%
	Done and BD not detected	97	35.14%
	Not done	73	26.45%
History of abortion in mothers	1	79	28.62%
	2 and more	20	7.25%
	None	177	64.13%

Table 2: Distribution of patients with respect to patients' birth weight and siblings with birth defect (n=276)

Birth weight	Low birth weight	115	41.67%
	Normal birth weight	151	54.71%
	Birth weight >4kg	3	1.09%
	Weight not known	7	2.53%
Siblings with BD	Yes and similar BD	7	2.53%
	Yes and dissimilar BD	36	13.04%
	No	233	84.42%

One hundred and fourteen cases (41.3%) had multiple birth defects and 162 cases (58.7%) had isolated birth defects. Cases of isolated BDs (n=162) most commonly involved musculoskeletal system

(41.36%) followed by cardiovascular system (24.08%) and central nervous system (22.22%). Details of isolated BDs, multiple BDs and systems involved in isolated BDs are shown in Fig. 1 and Figure 2.

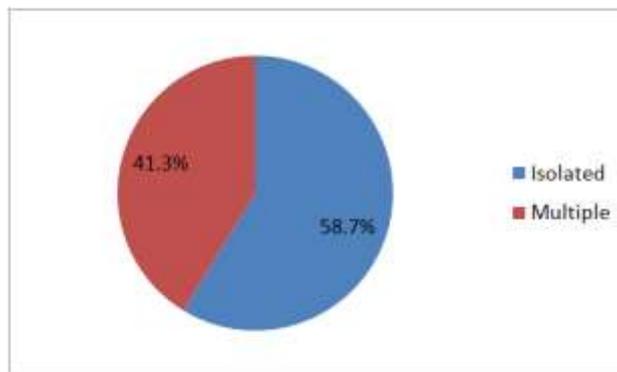


Fig. 1: Distribution of birth defects as isolated and multiple birth defects (n=276)

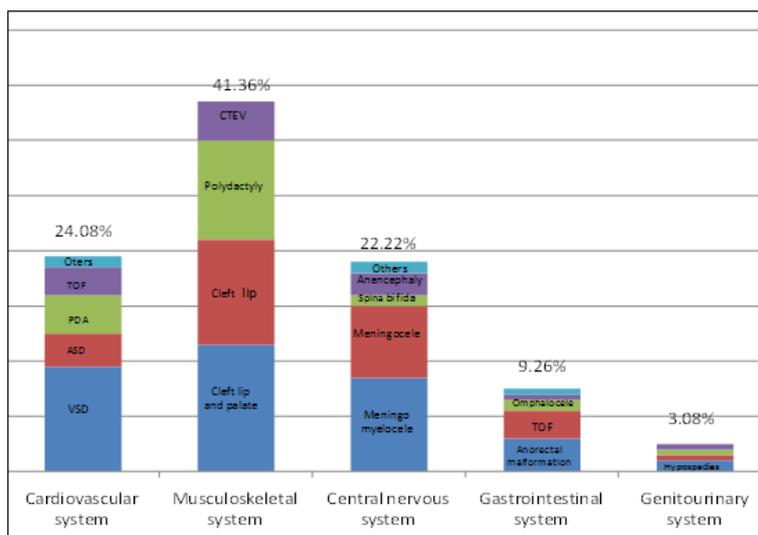


Fig. 2: Systemise distribution of isolated birth defects (n=162).

DISCUSSION

The mean maternal age at conception in the present study was 27.2 years (age range of 16 to 38 years and standard deviation of 4.2 years. Marlene A [6] has reported that there is higher prevalence of birth defects and chromosomal defects among mothers older than age 35. Yang JH *et al.* [7] stated that the mean maternal age is 29.9 years and 19% of the birth defects were associated with an elderly mother (≥ 35 years). Shi LM *et al.* [8] observed that the mean maternal age is 30.3 years for all live births, with 30.3 years for non-defects and 30.7 years for defects. Patel *et al.* [9] and Swain *et al.* [10] have shown higher incidence of malformations in the babies born to mothers aged over 35 years. Since the present study was conducted in a government district hospital, maximum number of mothers would be belonging to lower socioeconomic classes. Rates of early marriages, early conceptions, low education status and less awareness would be more and so the younger age of mothers.

Primigravida mothers constituted 33.69% of cases followed by 2nd (26.81%) and 3rd (22.83%) gravida in the present study. Kumar *et al.* [11], Aggarwal *et al.* [12], Yang JH *et al.* [7], and Shi LM *et al.* [8] observed that majority of the cases with malformations were

noted in the firstborn baby and showed a decline with increasing birth order. Swain *et al.* [10] and Tan *et al.* [13] reported that the prevalence of birth defect increased with birth order. Temtamy *et al.* [14] found no significant relation between birth order and the prevalence of malformations. Troung Hoang et al showed an increased prevalence of external birth defects (EBD) occurring among mothers with either primigravida or gravida over 4. The relationship between the mother’s age at delivery and gravidity may be one possible explanation for the high rate of EBDs at both extremes of maternal gravidity [15].

In the present study, BDs were detected antenatally in only 38.45% of cases, whereas ultrasound was not done in one fourth of cases. According to Ewigman *et al.* [16], the sensitivity of ultrasound in detecting congenital anomalies varied widely depending upon the age of the fetus, the skills and training of the person performing the exam, the time devoted to the exam, the type of anomaly, and the organ system affected, among other factors. Anderson *et al.* [17] estimated that overall sensitivities were much higher for central nervous system anomalies than for cardiac, skeletal, and craniofacial anomalies.

In the present study, mothers of 35.87% (n=276) of cases had history of abortions and mothers of 7.25% (n=276) of cases had history of multiple abortions. Thong MK *et al.* [18] had stated that mothers with affected babies were associated with significantly higher rates of previous abortions. Tan KH *et al.* [13] had mentioned that there was an increasing trend of abortion for birth defects, accompanied by a falling trend in the congenital anomalies of live births. Both extremes of maternal age were at higher risk of non-chromosomal birth defects while advanced maternal age was at higher risk of chromosomal defects.

In the present study, 15.57% (n=276) had siblings with BD. The rate of dissimilar BD (13.04%, n=276) is more compared to similar BD (2.53%, n=276). Rolv Terje Lie *et al.* [19] stated that among women whose first infant has a birth defect, the risk of the same defect in the second infant is substantially increased and the risk of a different defect in the second infant is slightly increased. Environment plays a strong part in repeated defects. The risk of having an infant with a birth defect varies among women. This heterogeneity is expressed in the relatively high risk of having a second infant with the same defect as the first infant. For example, among women who have one infant with a cleft lip, the risk of the same defect in the next infant is about 4 percent, [20] which is about 30 times higher than the risk in the general population.

In the present study 54.71% of cases had normal birth weight and 41.67% of cases had low birth weight. Marlene A [6], Patel *et al.* [9], Swain *et al.* [10] and Truong Hoang *et al.* [15] demonstrated that birth defects are significantly associated with preterm birth and low birth weight. Although preterm and low birth weight infants are more likely to have birth defects, the effect of birth defects on preterm birth and low birth weight has been difficult to study because of multiple confounding risk factors [8, 21]. Since the present study had included all paediatric patients, only those defects which are compatible with life and which would not have much effect on birth weight would be included more so the higher rate of normal birth weight.

In the present study 41.3% of cases had multiple birth defects and 58.7% of cases had isolated birth defects. Thong MK *et al.* [18] reported that 31.62% of cases had multiple birth defects and 68.38% of cases had isolated birth defects. Yang JH *et al.* [7] stated that multiple anomalies accounted for 16% of all birth defect cases. The rate of multiple BD is relatively high. So in any case of BD Physician should thoroughly search for other BDs.

Maximum number of cases of isolated BDs in the present study involved musculoskeletal system (41.36%) followed by cardiovascular system (24.08%) and central nervous system (22.22%). Marlene A [6], Shi LM *et al.* [8], Yang JH *et al.* [7] and Kumar *et al.*

[11] had observed that cardiovascular birth defects were the most commonly occurring birth defects, whereas Swain *et al.* [10], Verma *et al.* [22] and Temtamy *et al.* [14] had observed that the central nervous system was the most common system involved. Vishnu *et al.* [23] had documented that the musculoskeletal system was the most commonly affected.

CONCLUSION

Birth defects were more common in primigravida with the age group of 25 – 30 years. Not all the birth defects were detected antenatally. Whenever there is history of a previous baby with birth defect is present, the obstetrician or the treating physician should become very cautious for the next baby. Mothers of patients with birth defects had significant rates of previous abortions. The rate of having dissimilar BD in the siblings was more than similar birth defect. Birth defect cases more commonly had normal birth weight. Isolated birth defects were more common than multiple BDs but the rate of multiple BDs was significantly high. So whenever a birth defect is present one should search for other birth defects. The musculoskeletal system was the most common system involved in cases of isolated BDs.

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