

STUDY OF BIRTH DEFECTS IN A TERTIARY CARE CENTRE

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ABSTRACT

BACKGROUND

Birth defects are responsible for increased perinatal mortality and long-term morbidities. To reduce its incidence, which is the need of the hour we should know more about them and possible risk factors, which can be prevented.

The aim of the study is to study the overall frequency of birth defects in a tertiary hospital and search for association with certain risk factors.

MATERIALS AND METHODS

All newborns/stillborns with birth defects during one year were enrolled for the study. Similar number of newborns without birth defect during this period was taken as control. Relevant information was documented in both the groups and analysed.

RESULTS

Out of 11,008 births, congenital anomaly was found in 130 cases. The prevalence of birth defects was 1.18 percent. Association of occurrence of birth defects with increased paternal age, consanguinity, fever and drug intake in first trimester was found. 57.6% of the newborns with birth defects were stillborn, born at an earlier gestational age (33.6 week v/s. 37.5 weeks). Commonest system to be affected was CNS (49.2%).

CONCLUSION

Screening for aneuploidy and birth defects should be universal. Routine folic acid supplementation and pregnancy termination of malformed babies will reduce the incidences.

KEYWORDS

Newborn, Birth Defect, Neural Tube Defect, Consanguinity.

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BACKGROUND

Advances in medicine have led to decline in diseases like infection and malnutrition. Hence, congenital malformations have become important causes of perinatal mortalities and long-term morbidities.¹ Birth defects include abnormalities in the newborn baby's structure, function or body metabolism, which usually lead to physical and mental disabilities and can even be fatal sometimes.² The annual report of ICMR says that the commonest malformations are cardiac in nature. Cardiovascular, musculoskeletal and genitourinary were the most commonly affected systems in a descending order of frequency.³ But, according to the Birth Defects Registry of

India (BDRI), most common anomaly is the neural tube defect. Indeed due to large number of births per year (almost 27 million), India has the largest number of birth defects in the world. Birth defects affect approximately 1 in 33 infants and result in approximately 3.2 million birth defect related disabilities per year.⁴

Congenital malformation ranks the third most frequent cause of perinatal mortality in India.⁵ Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. It has been found that there is higher incidence of anomalies in stillborns and the incidence of congenital anomalies is significantly higher in preterm babies,⁶ low birth weight infants,⁷ maternal age above 35 years^{8,9} and increased birth order.^{6,7,8,9}

The accepted risk factors for birth defects are hydramnios, maternal febrile illness in the first trimester, past history of abortions, diabetes, eclampsia, history of congenital malformations in previous babies, parental consanguinity, hypertension and hypothyroidism.

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AIMS AND OBJECTIVES

The present study was conducted to know the incidence of recognisable malformations in the newborns and its association with different risk factors, so that this data will help the doctors and policy makers to plan effective prevention and intervention programmes.

MATERIALS AND METHODS

The present study was carried out in the Department of Obstetrics and Gynaecology of Sriram Chandra Bhanj Medical College, Cuttack, from May 2015 to April 2016. It is a prospective analytical study. All newborns and stillborns with birth defects, weighing more than 500 gm in labour room during this period were taken as cases. A detailed history was taken from the mother regarding age (both mother and father), literacy, socioeconomic status, locality from where she came and consanguinity of marriage, whether present or not. Enquiry was also made regarding periconceptional folic acid intake, febrile illness, infection or any drug intake during first trimester. She was interrogated about past pregnancy mishaps or history of delivering a malformed baby. Past history about diabetes mellitus and hypertension was asked. In current pregnancy, gestational age at delivery was calculated from LMP/early pregnancy USG and birth rate was taken. In stillborn, foetuses without obvious abnormalities, autopsy was done with parent's consent, and if malformation was detected, they were also enrolled as cases. In this way, 130 cases were taken. After delivery of each case, another newborn without any birth defect and delivered on the same day is taken as control. Similar numbers of normal newborns were enrolled as controls. The relevant data of controls were also taken.

RESULTS

Sl. No.	Characteristics	Case (n=130)	Control (n=130)	P value
1	Mean age	25.76	25.92	NS
2	Paternal age	31.67	29.73	<0.05
3	Mean GA	33.67	37.53	<0.01
4	SE status	Low=111, Medium=17, High=2	Low=120, Medium=9, High=1	0.27
5	Habitat	Rural=117, Urban=13	Rural=120, Urban=10	0.79
6	Consanguinity	7	0	0.007

Table 1. Sociodemographic and Economic Characteristics

Table 1 depicts mean maternal age of cases and controls were 25.76 and 25.92, respectively. But, mean paternal age of cases was 31.67 years where as that of controls was 29.73, which was statistically significant. There was no statistically significant difference in socioeconomic status and habitat (rural/urban) between both the groups. But, consanguinity of marriage was seen in 7 cases whereas no history of consanguinity of marriage was seen in controls, which was statistically significant (p=0.007).

Sl. No.	Characteristic	Case (n=130)	Control (n=130)	p value
1	h/o birth defect	2	0	0.156
2	h/o stillbirth	2	1	0.561
3	h/o spontaneous abortion	7	13	0.163
4	Past h/o diabetes mellitus	2	1	NS
5	Past h/o hypertension	1	1	NS
6	No bad obstetric history	116	114	-
	Total	130	130	-

Table 2. Past Obstetrics History

Table 2 analyses the past obstetrics history like history of previous birth defects, stillbirths and spontaneous abortions. There was no statistically significant difference between the two groups. Two mothers from case group and one from control group were known cases of diabetes mellitus. One mother from both the groups were known cases of chronic hypertension, hence the difference was not statistically significant.

Sl. No.	Characteristic	Case (n=130)	Control (n=130)	P value
1	Periconceptional folic acid intake	0	2	NS
2	h/o fever in 1st trimester	10	0	0.001
3	h/o drug intake in 1st trimester	6	0	0.013
4	Gestational age	33.67	37.53	<0.01
5	Pregnancy outcome	Live=55 Stillbirth=75	Live=130 Stillbirth=0	<0.05
6	Gender of baby	Male=61 Female=69	Male=63 Female=67	0.8

Table 3. Relevant History in Present Pregnancy

Table 3 shows all mothers of case group and 128 mothers of control group have not taken periconceptional folic acid. Ten mothers of case group gave history of fever in 1st trimester whereas no mother of control group gave such history. This difference was statistically significant. Mean gestational age at delivery were 33.67 weeks and 37.53 weeks in cases and controls respectively, which was statistically significant difference. Out of 130 cases, 75 were stillborn whereas no stillborn was taken in control group. There was no statistically significant difference between cases and controls when gender is considered.

Sl. No.	Type of Birth Defect	Frequency N=130	Percent
1	CNS (neural tube defect)	61	46.92
2	CVS	1	0.77
3	GIT	21	16.15
4	Musculoskeletal	40	30.77
5	Genitourinary	6	4.62
6	Orofacial	1	0.77
	Total	130	100

Table 4. Type of Birth Defect (in Cases Only, n=130)

Table 4 depicts most common malformation seen in our study was CNS malformation, which was seen in 61 cases (46.92%). Musculoskeletal malformations rank next, that is 40 cases (30.77%). GI malformations were seen in 21 cases (16.15%). Only 1 case (0.77%) of CVS malformation was noted in our study.

Urogenital malformation was seen in 6 cases. (4.62%). Orofacial (bilateral cleft lip) was seen in 1 (0.77%) case.

DISCUSSION

Prevalence of birth defect in our study was 1.18 percent. Similar result was reported by Patel Z.M. and Adhia R.A., which was 1.63 percent.¹ But, according to global statistics, the incidence of birth defect is around 2-3%.¹⁰ The low incidence in our study maybe due to lack of routine TIFA scan and examination of newborn by neonatologist. Newborns with internal malformations were missed who did not manifest in early neonatal period.

The paternal age effect was first proposed implicitly by Weinberg in 1912¹⁰ and explicitly by Penrose in 1955.¹¹ But, in our study, association between higher paternal age and birth defect was noted.

Consanguinity was found in 7 cases (5.4%) in our study, but Patel ZM, Adhia RA found consanguinity in 8.1% of cases.¹

In present study, history of fever in 1st trimester was found in 7.7% of cases, which is higher than a Chinese study by Zhiwen Li, Aiguo Ren.¹²

In our study, 6 cases (4.6%) had history of drug intake in 1st trimester (category C and D drugs).

The mean age of delivery in our cases was 33.67 weeks. A similar study was conducted by Patel ZM, Adhia RA where they found increased incidence of delivery at lesser gestational age with malformations.¹

In our study, out of 130 malformed babies, 37.6% were stillborns. In the study of Shamnas M et al congenital malformation contributes to 19.5% of perinatal mortality and 9.9% of stillbirths.²

In present study, the commonest birth defect found was CNS (49.2%). In the study by Patel and Adhia, they have also seen this system to be most commonly affected among malformed babies. According to Birth Defect Register of India (BDRI) most common anomaly is neural tube defect. The annual report of ICMR says that the commonest congenital malformations are cardiac in nature.³ A cross-

sectional study by Abed Yehia et al reveals cardiac defects to be the most common birth defect, i.e. 45% in their study.¹³ Cardiac malformations are not externally visible, hence those newborns who manifest symptoms in the early neonatal period are only included in our study. So, this might be the reason for under reporting of cardiovascular malformations.

CONCLUSION

The incidence of neural tube defects has markedly reduced in the developed countries following mass promotion and mandatory prescription of folic acid for pregnant mothers. But, in developing countries like India, the incidence of neural tube defects is still very high, highest in many studies including the present one. The risk factors should be minimised and periconceptional folic acid intake should be made universal in our country to reduce the incidence of birth defects. With greater emphasis on small family norms and population control, emphasis should be for the early diagnosis of malformations. Screening should be universal and should start in first trimester both by biochemical tests and ultrasound.

Compliance with ethical requirements and conflict of interest.

Ethical approval was taken from Institutional Ethics Committee for this study (image included at the end of the document). Written and informed consents were taken from all the parents whose fetuses had undergone autopsy. Authors have no conflicts of interest relevant to this article.

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