A Study of Anatomical Congenital Anomalies in Newborns in a Teaching Hospital of India

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Abstract

Background and Introduction: Congenital malformations may be defined as any anatomical defect, single or multiple, present at birth, attributable to fault in development. The severity of such a defect may vary from symptomless and regressible capillary naevi to lethal multiple system defects. A combination of genetic, biologic and environmental factors is considered to be responsible for many of these conditions. *Materials and Methods:* The study was carried out in the Obstetrics & Gynecology department of a tertiary care hospital in Southeastern India. The period of study was taken as per the protocol of the project. It was a hospital based observational study not involving any surgical intervention. *Observation and Results:* A total of 7268 babies were delivered during this period in the Department of Obstetrics and Gynecology, (both live and still born). Out of these 116 (one hundred and sixteen) babies were found to have congenital malformations including both major and minor malformations. In the present study, the commonest system involved was the musculoskeletal system (22.4%). The commonest malformations of the musculoskeletal system observed in the present study were talipes (15 cases) and polydactyly (8 cases). *Conclusion:* Congenital malformations constitute one of the important causes of morbidity and mortality in the neonatal period, affecting nearly 1.59% of the newborns and contributing to many of the prenatal deaths.

Keywords: Congenital; Malformations; Neonates; Defects; Newborns.

Introduction

Advances in medicine have led to decline in diseases like infection and malnutrition. In The present scenario, congenital malformations have emerged gaining great importance in perinatal mortality [1]. Congenital malformations represent defects in morphogenesis during early fetal life. It is recognized that maldevelopment is the result of subtle interplay between gene and environment. Also it has been observed that better maternal care and improved standards of living have very little effect on overall frequency of congenital malformations [2].

Congenital malformations affect 2.5% infants at birth and are responsible for about 15% perinatal mortality in India [3]. According to some authors [4], the incidence of congenital malformations is approximately 2% of total births. A biologic phenomenon which has never been explained is noted by the fact that the incidence of congenital malformations is similar for all populations although the frequency of specific malformation differs from country to country [5].

The causes of majority of congenital anomalies are not currently understood. A combination of genetic, biologic and environmental factors is considered to be responsible for many of these conditions. However, the cause is not known in about 40-60% of cases [6].With all this in view the present study was undertaken to evaluate a spectrum of congenital malformations in a teaching hospital in southeastern India.

Materials and Methods

The present study was conducted in the Department of Obstetrics and Gynecology, in a teaching hospital in southeastern India. The study

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was done from July 2007 for a period of 3 years. A total of 7268 babies were delivered during this period (both live and still born). Out of these 116 (one hundred and sixteen) babies were found to have congenital malformations which included both major and minor malformations

Babies were examined within the first 3 days of delivery for congenital malformations. All live born babies born during this period and stillborns after 28 weeks of gestation or whose weight was about 1 kg were included in the study. Products of conception expelled by mid-trimester, abortion were not included in the study. A thorough physical examination was done within 24 hours of delivery. The live borns with congenital malformations, who survived at the time of discharge were again examined at 3 days of life and prior to discharge and were followed up when possible. The still borns were autopsied as and when consent from parents was obtained. Despite best efforts few still borns could not be studied properly due to lack of consent for autopsy by parents.

Every newborn baby was subjected to a detailed examination from head to toe within the first 24 hours of birth and again at 3 days of life. All macroscopic anatomical defects detected were recorded in a pre-designed Performa. Major congenital malformations included those defects which caused serious structural, cosmetic and functional disability and required surgical or medical intervention. Minor malformations were defined as those that were not lethal like skin tags, capillary hemangioma, preauricular sinuses, nevi, accessory nipples, tongue-tie, congenital teeth etc. Although these were not so important, the presence of these minor abnormalities often alerted for undertaking a more thorough clinical search to unearth associated major malformations.

Congenital malformation like pyloric stenosis could not be studied as it does not present within 7 days of life. The clinical diagnosis was complemented by investigations as and when necessary like radiological investigations. Chromosome studies could not be done. Only gross anatomical study was done. Histopathological study was not done.

Ethical Statement

Ethical clearance was taken from the institutional ethical committee. Consent forms were duly completed by the parents and the only the cases were documented. It was an observational study done in hospital settings.

Statistical Analysis

The data was collected and tabulated. They were then statistically analyzed by the ANOVA test and Probability test [7]. The software used was MSTATC. The website referred was www. graphpad.com.

1	Baby of _____	7	Weight_____
2	Regd No_____	8	Single or twin_____
3	Sex of Baby _____	9	Live or still born_____
4	Date & time of birth_____	10	Season of the year____
5	Date of Examination_____	11	Gestational age _____
6	Mode of Delivery_____		

1. **Examination of various systems: -**
   A. CNS and Spine
   B. CVS
   C. Alimentary system
   D. Genitourinary system
   E. Respiratory system
   F. Musculoskeletal system
   G. Orofacial (including ocular, auricular, nasopharynx, palate, nose, lip)
   H. Skin
   I. Others

2. Baby is having single cong./Multiple congenital anomalies.

3. Investigation done in live born.

4. Whether baby survived the first 7 days of life

5. Autopsy finding in still born.

Observations

The present study was carried out in the above teaching hospital in Southeastern India for a period of three years. A total of one hundred sixteen (116) babies were (Table 1) found to have congenital malformations. During this period 7258 women delivered 7268 babies. Ten pairs of twins were delivered. Out of the twins, two babies had congenital malformations.

Table 1: Shows incidence of congenital malformation during the period of study

		Total cases	Malformed No.	Percentage
1.	Total births	7268	116	1.59
2.	Live births	7108	100	1.4
3.	Still births	160	16	10.0*

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Central Nervous defects were seen in 31 babies. Hydrocephalus was the commonest anomaly observed having an incidence of 1.4/1000. Sacrococcygeal teratoma and facial palsy each had an incidence of 0.3/1000.Among babies with nervous defects 8 had anencephaly and were born dead.

Musculoskeletal defects were seen in 32 babies. Talipes equinovarus was most common and seen in 15 cases. Polydactyly was the 2nd most common abnormality and was seen in babies with other associated defects. 25 babies showed orofacial defects out of which cleft lip with cleft palate was seen in 16 with an incidence of 2.2/1000. Both hypo plastic nose and pre-auricular skin tags showed an incidence of 0.14/1000. Gastrointestinal defects were seen in 6 babies in our study. Out of them 1 rare case of gastrointestinal atresia having esophageal and duodenal atresia was also observed.

3 babies were born with congenital varicella and had vesicles all over the body. Urogenital defects were observed in 21 babies with as many as 7 showing hypospadias.

An interesting case of conjoined twins was also reported having a fused heart. It was a case of thoracopagus and one of them had cleft lip. They had a single anterior placenta with a succenturiate lobe. Cardiovascular defects as they do not present within 3 days of delivery were not commonly seen within our observation period. Table 2 shows the percentage incidence of malformations of various systems, in relation to total number of babies with malformations. In this table, individuals with malformation of a single system are entered for that particular system. Those with multiple malformations are grouped together in a single group of multiple malformations.

Table 2: Shows the percentage incidence of malformations of various systems, in relation to total number of babies with malformations

Sl. No.	System	No. of babies	% of total malformed babies
1.	CNS	21	18.1
2	Musculoskeletal	26	22.4
3	Orofacial	19	16.4
4	Ear	3	2.6
5	Eye	2	1.7
6	GIT	4	3.4
7	Urogenital	19	16.4
8	Skin	6	5.2
9	CVS	0	0
10	Down's syndrome	4	3.4
11	Multi system involvement	12	10.4
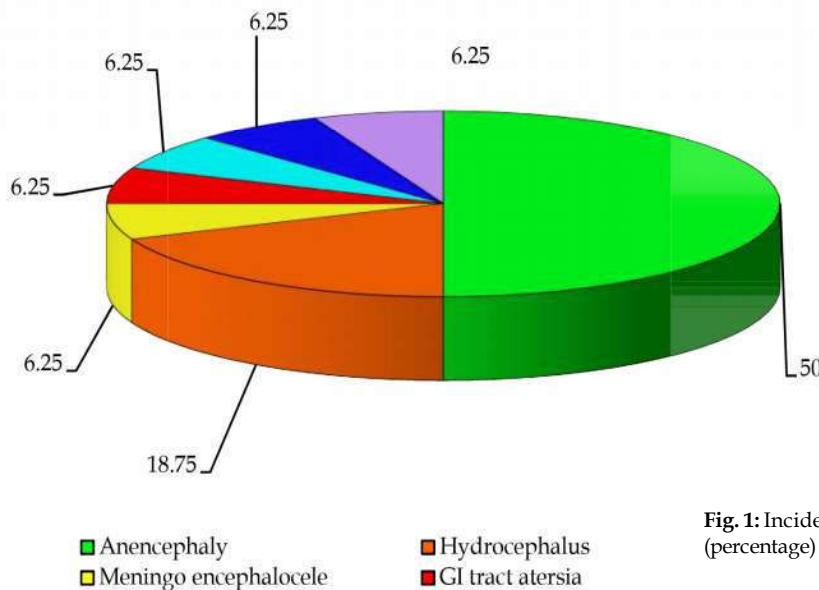
Fig. 1: Incidence of major malformations (percentage) of total malformed still borns

In this period of study, a total of 160 babies were still born of which16 were malformed. From Fig. 1, it is seen that 81.25 % of still born infants had congenital malformations of the central nervous system, the incidence of which is quite high when compared with incidence of CNS.

Malformations of total malformed babies which was 18.1% (p value<0.01). 72 babies showed major malformations and 44 had minor malformations.

Table 3: Shows the distribution of male and female babies born with congenital malformations

Sl. No.	Total cases	No	Percentage
1.	Male	68	58.6
2	Female	48	41.4

As seen in above table the % incidence of male malformed babies is 58.6 while (Table 3) that of females is 41.4(p<0.05). M:F ratio is 1.4:1 which is significant. There were no babies below 1 kg because midtrimester abortions were not included in our study. Majority of malformed babies (72.4%) were low birth weight; their weight ranging from 1-2.5 kg. Malformations were seen only in 10 babies with birth weight>3 kg (8.6%). In our period of study 10 pairs of twins were delivered out of which 2 had some or other malformations accounting for 10% which is significantly high compared to single births where it was 1.57% (p<0.05%). 18 babies died during their hospital stay of 3 days. Among these 10 died due to (Table 4) malformations and 8 died due to extreme prematurity. Also, surgical intervention was done in a few cases and was successful.

Table 4: Shows the number of deaths among the babies born with malformations

Sl. No.	Malformation	No. of deaths
1	Conjoint twin with fused heart	1
2	Hydrocephalus	6
3	Encephalocele	1
4	Fetal ascitis	1
5	Gastroschisis	1
	Total	10

* Total live born 100

Discussion

The present study revealed that the incidence of congenital malformation per thousand total birth is 15.9/1000 or 1.59%. The incidence in different studies can vary depending upon the source of

malformation, population sampled, selection of study material, astuteness of clinician and availability of laboratory aids. In fact, only 43% of malformations could be diagnosed at birth as reported by some authors [8]. The incidence in the present study which was found to be 15.9/1000 births agrees with authors [9] from other countries who found it to be 13.5/1000 and authors from north India [10] who observed 20/1000. Chinara from BHU [11] observed a gradual decline of incidence of congenital malformation from west to east India (Figure 2 and 3) from about 3.6 in Chandigarh to about 0.2 in Kolkata.

Fig. 2: A baby with Gastroschisis

Fig. 3: A baby showing congenital varicella

Fig. 4: A newborn baby with accessory nipples

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It has also been noted that congenital malformation in a birth cohort in an urban population increase from 26.22/1000 to 40.37/1000 at 5 years. This occurs because many malformations present themselves in late childhood [12]. In the present study, the commonest system involved was the (Figure 4) musculoskeletal system (22.4%). This system as the commonest system involved was also noted in other studies [13] conducted by the team of Chaturvedi et al.

Malformations of the central nervous system were reported to be commonest by most of the Indian researchers and workers. The incidence of multiple malformation involving different systems was found to be 10.4%. In our study out of 12 cases of multisystem involvement, CNS malformations presented as the principal malformation [14]. Authors across the world have found central nervous system abnormalities in 80% of stillborn malformed babies. The relative higher incidence of neural tube defects in our study (2.3/1000) is in keeping with few other north Indian studies [15-16]. Malformation of the orofacial area (excluding eye and ear) was found to have a high incidence (16.4%) in the present study. Mathur et al. [17] reported an incidence of 3.8/1000 for cleft lip (Figure 5) and cleft palate.

Fig. 5: A baby with Cleft lip, cleft Palate and anencephaly

Malformations of the gastrointestinal system in the present study accounted for only 3.4% of cases. However, malformation of this system was found to be commonest by some authors in community-based studies [18]. In the abnormalities of the genitourinary system (16.4%), hypospadias was found to be the commonest (0.96/1000). Hypospadias was also reported to be the commonest anomaly by one study by Mittal et al. [19]. The incidence of cardiovascular system anomaly is low at birth relatively because many are not detected till school age.

There were 4 babies born with Down's syndrome showing typical features. If we compare the still born children in our study it was 2.3% whereas other reports show it 3.7%, 4.4% and 2.4% in various other studies. A total of 72.4% malformed babies were low birth weight. It has been noted that 70% of infants with internal anomalies had birth weight less than 50th percentile for their gestational age [20-21].

Conclusion

So, to conclude congenital malformations constitute one of the important causes of morbidity and mortality in the neonatal period, affecting nearly 1.59% of the newborns and contributing to many of prenatal deaths.

One of the major steps in reducing the incidence and proper management would be early detection. Medical termination of pregnancy for uncorrectable severe malformation should be done and provision of antenatal diagnosis by use of ultrasonography, amniotic fluid studies and other methods are important.

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Conflict of Interest: None declared

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