

STUDY OF GROSS CONGENITAL MALFORMATIONS IN NEWBORN

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ABSTRACT: In First Trimester, organs of various systems in body of fetus develop, the process known as "Organogenesis". Any interruption in this process of organogenesis will lead to defective formation of that particular affected organ. The factors like genetic, environmental, teratogenic and infectious agent play an important role for the origin of malformation during the most sensitive period of embryogenesis. With Advances in Medical Research, we can diagnose these malformations at an earlier gestational age. There are various modalities in form of invasive and non-invasive procedures in first and second trimesters to diagnose and to treat these malformations to reduce incidence of malformations, as well as to reduce the turmoil of parents. This study is conducted to deduce incidence of Congenital Malformations in newborn and fetus, delivered in our institute. A retrospective study of 100 cases of gross congenital malformations in newborn which presented in Sheth L.G. Hospital was carried out from October 2008 to August 2010. The majority of malformations noted in this study belonged to central nervous system (36%). The second most commonly involved system was musculoskeletal system (23%). Gastrointestinal system is the third most commonly involved system in this study (13%).

KEY WORDS: Gross congenital malformation, central nervous system, musculoskeletal system, prematurity, stillbirth

INTRODUCTION: Genetic and congenital diseases are almost always serious, incurable, a number of these diseases are treatable, and in some cases, their clinical therapeutic intervention and study of family history and genetic counseling remains of paramount importance. Genetic and congenital abnormalities are more than what is generally appreciated and are the cause of significant morbidity and mortality in pediatrics. With decreasing incidence of fatal infectious diseases, congenital anomalies would be one of the main causes of infant's mortality in future. Hence, we investigated large heterogeneous populations of paediatric patients to determine the current prevalence of genetic and congenital anomalies among our populations. Congenital Malformations has defied fully satisfactory solutions till this day, even in present atomic age. The incidence of congenital malformations varies from country to country and from one region to another in the same country. The aim of this study is to determine the pattern of congenital anomalies and to establish any inter-relationship. Congenital anomaly remains one of the leading causes of Infant mortality. Thus the place of this research in the programming of a better health care delivery cannot be overemphasized.

METHODS: The study conducted was within time period of 23 months from October 2008 to August 2010. Antenatal History included Maternal Age, Parity, Consanguinity, and Gestational Age at the time of Diagnosis, History of Drug intake, Associated Maternal Conditions, Previous History of affected children. All live birth and still born babies were examined for the presence of congenital malformation at the time of birth. Detailed general and systemic examination of the baby was

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carried out. Baby's gestational age and birth weight were noted. In antenatal diagnosis, 2-D USG was used as diagnostic modality in most cases because of ready availability and low cost. X-ray was used to diagnose mainly skeletal disorders in postnatal babies only to avoid intrauterine radiation exposure. Due to financial constraints, non cooperation and lack of easy availability, all the investigations could not be carried out in all of the patients.

RESULTS: Out of total 8257 deliveries from October 2008 to August 2010, gross congenital malformations were noted in 100 newborns, making it an incidence on 1.21%. Out of the congenital malformations noted, the most commonly involved system was central nervous system with 36 cases (36%), followed by musculoskeletal system with 23 cases (23%), Gastrointestinal system with 13 cases (13%), Genitourinary system with 11 cases (11%), cardiac cases with 4 cases (4%), Vascular & lymphatic with 6 cases (6%), multisystem involvement in 5 cases (5%), Ear Nose Throat involvement in 2 cases (2%). Out of total live births (7945), congenital malformations were seen in 58 cases (0.73%), whereas, out of total stillbirth (312), congenital malformations were seen in 42 cases (13.46%). Out of 100 congenitally malformed neonates' deliveries, only 38 occurred at full term (38%), whereas 62 were preterm (62%).

DISCUSSION: The incidence of congenital malformation in present study is 1.21%. This was comparable to the studies conducted by some other Indian Authors^{1,2,3,4}.

The incidence was comparable with the incidence in other countries like China (1.1%)⁵, Russia (1.23%)⁶, UAE (1.5%)⁷. There were increased incidence rates noted in England (2%)⁸ & United states (2-3%)⁹. These variations might be explained by social and racial influences which are commonly known in genetic disorders.

Central Nervous System was the most commonly involved system (36%), comparable with studies by Neelam and Grover et al (2000)⁴, Tuncbilek et al (1999)¹⁰. (Table 1) In central nervous system, Anencephaly (7 cases), Meningocele (7 cases) and hydrocephalus (6 cases) were most commonly noted.

The second most commonly involved system was musculoskeletal system (23%), of which cleft lip and cleft palate (6 cases) and Talipes equino varus (4 cases) were common.

In Gastrointestinal system (13%), tracheoesophageal fistula, congenital diaphragmatic hernia, duodenal atresia, omphalocele were common.

Bilateral multicystic Dysplastic Kidney was commonest amongst genitourinary system.

Out of 100 cases, there were 2 cases of Down's syndrome, 1 case each of Pierre Robin Syndrome, Golden Harr Syndrome, Backwith Widman Syndrome.

Out of total still births, 13.46 % had congenital malformations, is higher than the rate among the live births. The cause of still birth in those cases was congenital malformation in nearly 50% but certain other maternal factors were also responsible (Table 2). This was comparable with studies by other Indian Authors^{1,4}.

Congenital malformations were observed in preterm neonates more frequently than in full term neonates (Table 3). Congenital anomalies and syndromes are associated with premature labor. In fact, many of these fetuses are spontaneously aborted very early in pregnancy^{11,12}. Of those who are carried beyond the first half of pregnancy, more than half are delivered preterm^{13,14}, and they

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may have restricted intrauterine growth¹⁵. Some anomalies and syndromes are associated with both preterm delivery and intrauterine growth restriction.

System	No. of cases	Percentage
Central Nervous System	36	36%
Musculoskeletal	23	23%
Gastrointestinal	13	13%
Genitourinary	11	11%
Cardiac cases	4	4%
Multisystem	5	5%
Vascular and lymphatic	6	6%
ENT	2	2%

TABLE-1: CONGENITAL MALFORMATION ACCORDING TO VARIOUS SYSTEMS

	Total cases	Number of Anomalous fetus	Percentage
Total Births	8257	100	1.21 %
SB/IUD	312	42	13.46 %
LB	7945	58	0.73 %

TABLE-2: STILL BIRTH AND CONGENITAL MALFORMATIONS

Maturity	Total deliveries	No. of cases	Percentage
Full term	7207	38	0.58%
Preterm	1010	62	6.14%

TABLE-3: PREMATURITY AND CONGENITAL MALFORMATION

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IMAGE 1: Cystic Hygroma



IMAGE 2: Meningocele



IMAGE 3: cleft lip and cleft palate



IMAGE 4: Alobar Holoprosencephaly

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