A Study of Congenital Anomalies In Newborn

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Abstract: Total 4210 babies were studied in the neonatal period immediately after birth, which shows overall 0.88% incidence of congenital malformations. Incidence was significantly higher (6.1%) in mothers aged > 30 years as compared to younger age group. Religious pattern of mother and consanguinity of marriage had no much effect on the incidence. The incidence was marginally higher in primiparous patients (47.2%). Only in 12% of cases some etiological factors could be found, of which maternal fever in first trimester was the most common. 10 cases with CNS malformations had hydroamnious. The anomaly was detected antenatally in 49% cases with the help of USG. The ration of still birth to live birth was 4.19:1 suggesting that many of the anomalies are incompatible with life. There was no overall difference in the sex of the babies. However most of anencephalic babies were female. Preterm babies had four times more incidence of congenital malformations as compared to term babies. This represents “phenomenon of nature’s selection”. Associated hydroamnios also accounts for preterm labour similarly congenital malformations were more commonly found in low birth weight baby. Out of all system involved in congenital malformation, CNS was the commonest and accounted for 1/3 rd of the cases.

Key Words: Congenital, Abnormalities, Neonates

INTRODUCTION: Early intrauterine period during 3rd – 8th weeks of gestation is the vital period of life for the normal development of organs and organ system or organogenesis. It was observed that better maternal care and improved standards of living have very little effect on the overall frequency of congenital malformations. 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; or defined more widely to include functional disturbance as a defect, any irreversible condition exiting 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.

A congenital anomaly is thus any alteration present at birth of normal anatomic structure and has cosmetic, medical or surgical significance. The birth of an infant with major malformations, whether diagnosed antenatally or not, evokes an emotional parental response.

Congenital malformation will begin to emerge as one of the major childhood health problems. Treatment and rehabilitation of children with congenital malformations is costly and complete recovery is usually impossible. Approximately, 66% of major malformations have no recognized etiology and most of them have multifactorial inheritance. These defects can occur for many reasons including inherited genetic conditions, poor diet, toxic exposure of the fetus for example, to alcohol, birth injury and, in many other cases, for unknown reasons.

Available literature shows that congenital malformations contribute highly to prenatal mortality and postnatal physical defects. Parents are likely to feel anxious and guilty on learning of the existence of a congenital anomaly and require sensitive counseling.

Written records of congenital malformations have come down to us from the ancient

The worldwide incidence of congenital disorder is estimated at 3-7%, but actual numbers vary widely between countries. Congenital malformations affect 2.5% of infants at birth and are responsible for about 15% of perinatal mortality in India.

The present study was carried out to determine the overall incidence, types and distribution of various congenital anomalies both in live born babies and still born babies, and also to find out the major associated maternal and fetal factors, which can help physician to identify the cases early. So that appropriate prophylactic measures can be taken in time, which will prevent handicaps resulting out of congenital anomalies, which will help our developing country to become healthier.

MATERIALS & METHODS: This was a descriptive, cross-sectional study of newborns and stillborn babies delivered at Sir T Hospital during period from January 2006 to June 2007. Data collection was performed by means of structured from which contained two parts. At first part, variables recorded were about maternal characters and included the date of admission, age, history of chronic illness, drug ingestion, exposure to X-ray, history of CM in other offspring, parental consanguinity, and were obtained by interviewing with neonates, mother. The second part was about neonatal characters including live, or stillbirth, gestational age, birth order, sex, existence of congenital anomaly and type of it. No autopsy examinations were performed.

Once the diagnosis of major malformation (incompatible with life) was confirmed, pregnancy was terminated by different methods of induction of labor. Mode of delivery was recorded in all Babies were examined jointly by anatomist, obstetrician and pediatrician. The birth weight, sex of baby and nature of anomaly was carefully noted. Some of babies were subjected to surgical correction of anomaly if needed in the immediate neonatal period. The mothers of affected babies were asked again about exposure to any probable etiological factors during pregnancy or positive family history. Observation was tabulated and analyzed at the department of Anatomy, Medical College, Bhavnagar.

RESULT: With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developed countries and would very soon be increasingly important determinants of perinatal mortality in developing countries like India.

The Study was carried out and observation was tabulated and analyzed at the department of Anatomy, Medical College, Bhavnagar. Out of 4098 singleton delivery there were 35 baby delivered with one of the malformation, we had 55 pairs of twins and 1 pair of triplets, and we had only 2 malformation among this group. Out of 2 twins’ deliveries, they have Dwarfism and Anencephaly with spina bifida respectively. In all cases second pair of twins babies were normal. Thus, A Total 4210 babies born to 4154 mothers, out of which 37 babies had some anomalies, which is approximately 0.88%.

Graph I

The incidence of anomalies was however markedly high (6.1%) in mothers with age > 30
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Yrs (shown in graph I) whereas others have associated 20-35 years maternal age group with higher incidence of congenital malformation24. Down’s syndrome is an important example of age related malformation.

Incidences of congenital malformation were slightly more in female with F: M ratio of 1.6: 1. Many studies have documented male preponderance amongst congenital malformed babies 23, 25 Religious pattern of mother and consanguinity of marriage had no much effect on the incidence.

In present study the incidence of congenital malformations declines with increasing birth orders. Amongst Malformed babies 42% were born to primigravida. The relative higher incidence of malformation in primigravida has also observed by Desai et al26 while Anand et al27 have found no significant correlation between birth order and congenital malformation.

### TABLE I : PATTERN OF MALFORMATIONS ACCORDING TO ANC

<table>
<thead>
<tr>
<th>ANC</th>
<th>Cases n = 4154</th>
<th>No of malformed based on n = 37</th>
<th>Incidence (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Booked</td>
<td>2361</td>
<td>9</td>
<td>0.36</td>
</tr>
<tr>
<td>Emergency</td>
<td>1793</td>
<td>28</td>
<td>1.50</td>
</tr>
</tbody>
</table>

The Table I shows that the incidence of malformations is slightly higher in those mothers who had not received antenatal care. Clinical suspicious supported by USG help in detection of some anomalies in booked cases. These patients are offered selective termination of pregnancy. Only in 12% cases etiological factors could be detected. Commonest being fever in 1st trimester. In remaining 88 %, we are not able to find out the cause of anomalies.

Anand et al27 have observed a statistically significant correlation between 1st trimester fever and positive familial history and congenital malformations. In present series 3 mothers had history of drug ingestion in early pregnancy mostly in the form of antibiotics an antiepileptic. However it was difficult to find exact nature of drug in all cases. Twins and radiations also accounted for small no of cases. In the present study also there is a striking correlation between hydromnios and neural tube defects. Out of 37 cases of congenital malformation we were able to detect only 54 cases antenatally.

Most of babies with congenital malformation delivered vaginally. Two patients required LSCS for hydrocephalus. One had emergency LSCS and diaphragmatic hernia diagnosed. One patient having multiple congenital anomalies and another having CTEV, they all were emergency patients

Congenital malformations were more in pre term deliveries. It is known that abnormal fetuses are likely to be delivered prematurely or aborted, based on the fact that a significant number of babies have chromosomal anomalies. This represents “phenomenon of nature’s selection”. Whether the relationship between pre maturity and congenital is cause or effect is difficult to comment.

In present study out of all malformed babies 79% were heaving low birth weight and there is inverse relationship between incidence of malformations and birth weight (shown in Graph II) This association of low birth weight and malformations has been well documented 23,28.

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CNS (64.56%) followed by GIT (14.17%). Amongst the CNS Malformations neural tube defects were the commonest of which anencephaly with or without spina bifida constituted ¾ of cases. Most of anencephalic babies were female (66.6%). Majority of CNS malformations were incompatible with life and 69.5% were still births. The GIT and Musculoskeletal system is next common system involved in congenital malformation and are surgically correctable.

Out of total 37 malformed baby, 2 babies were having multiple system abnormality and in 15 babies there were more than one system involved thus total 127 malformation were observed in present study. Mishra and Bhaveja reported multiple anomalies in 37.6% of anomalies.

A significantly higher incidence of malformation observed among the stillbirths (13.30%) in the present study as compare to live birth (3.2%). It is consistent with earlier reports. Aiyar and Agrawal observed that the highest incidence of malformations was among full term normal weight babies.

Thus, congenital malformations are emerging as important perinatal problem contributing to the perinatal mortality and morbidity. The difference between the frequency of types of congenital malformation in different parts of this country and reports from other countries may be due to genetic background and geographic nutritional and socioeconomic differences. More research is needed to determine the factors underlying the various types of congenital malformation encountered in this area.

CONCLUSION: The study definitely helps to know the pattern of congenital anomalies and the relationship of various gestational and familial factors in relation to congenital anomalies. Use of folic acid prior to and during first trimester can prevent neural tube defects. Malformation scan can detect lethal congenital anomalies. Antenatal testing like amniotic fluid testing can be used to detect certain lethal congenital anomalies, hence maternal education and family planning play a very important role in prevention of congenital anomalies. Consanguinity is associated with increased incidence of anomalies so it should be discouraged.

Congenital malformations also have implication in society as it leads to increase incidence of still birth and neonatal death. Inherited and chromosomal anomalies are associated with loss of physical or mental and intellectual abilities. Often emotion upset and social stigma to parents are beyond the limit of our imagination.

Genetic counseling play a vital role for the high risk parents. It provides information regarding various procedure and diagnostic technique, the risk an consequences of some of the procedure as well as about options available. Genetic counseling at different time period helps in reduction of congenital anomalies, morbidity and mortality resulting from these anomalies.

REFERENCE:
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21. Singh M. Hospital-based data on perinatal and neonatal mortality in India. Indian Pediatr 1986, 23: 579-584


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