

Research Article

Incidence and distribution of congenital malformations clinically detected at birth: a prospective study at tertiary care hospital

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ABSTRACT

Background: Congenital malformation represents defects in morphogenesis during early fetal life. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. The objective was to study overall and individual incidence and distribution of clinically detectable congenital malformations in newborns delivered at a tertiary hospital.

Methods: The present study is a prospective study of all the newborns delivered at Obstetrics and Gynecology Department, New Civil Hospital, Surat, Gujarat, India for a period of one year from 1st January 2010 to 31st December 2010. Total 5518 consecutive births including both live born babies and still born babies were examined after taking verbal and written consent of their mothers for a visible structural anomalies to determine the overall incidence and distribution of congenital malformations. Data were statistically analyzed using SPSS software (trial version).

Results: A total of 5518 babies were born out of which 75 were twins. Out of total 5518 newborns 5316 were live births and 202 were still births and out of 5316 live births 48 babies were malformed and out of 202 still births 20 babies were malformed. Total numbers of malformed babies were 68, so total point incidence of congenital anomalies turned out to be 1.23%. Out of total 5518 babies, 35 (0.63%) were having central nervous system malformations making its incidence of 6.34/1000 live births which turned out to be highest followed by gastrointestinal system (incidence of malformed babies: 5.44/1000 births) and genitor urinary system (incidence of malformed babies :1.09/1000 births). Commonest anomalies in central nervous system were meningomyelocele followed by hydrocephalus and anencephaly.

Conclusions: From present study we conclude that incidence of congenital anomalies of CNS was highest amongst all types of congenital anomalies (meningomyelocele being the commonest). More emphasis should be given on prevention by regular antenatal care and avoidance of known teratogens and probable teratogenic agents.

Keywords: Congenital anomalies, Congenital malformations, Still births, Birth defects, Prenatal diagnosis

INTRODUCTION

The most traumatic experience for a gravid woman, her husband and their family is, undoubtedly, the unheralded birth of deformed child, precipitating feeling of horror, inadequacy and failure in parents.¹

Congenital malformation represents defects in morphogenesis during early fetal life. According to the

World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.² The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.^{3,4}

Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the pediatrician. The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centers providing good neonatal care. Also, increased use of irradiation, alkylating agents, antimetabolites, self-drugging, smoking, alcohol consumption has contributed to increased incidence of congenital malformation.

It is estimated that 1 in 40 or 2.5% of newborns have a recognizable malformation or malformation at birth.⁵ In India with decreasing mortality due to infection and nutritional disorders, incidence in death due to congenital malformation are increasing.⁶ A study done at AIIMS show that congenital malformations contributed to 13.4% of perinatal deaths as compared to 9% a decade back.⁷ Major malformation accounts for 15% of neonatal death.⁸

Studies like present series and other ongoing multicentric study programmes, are expected to alert us with regard to new teratogen, better understanding of the epidemiological implications and thereby helping us in preventing the occurrence and better management of congenital malformation. This study has been conducted to throw light on overall and individual incidence and distribution of clinically detectable congenital malformations in newborns delivered at a tertiary hospital.

METHODS

The present study is a prospective study of all the newborns delivered at Obstetrics and Gynecology Department, New Civil Hospital, Surat, Gujarat, India for a period of one year from 1st January 2010 to 31st December 2010. Before conducting the study approval was obtained from institutional ethical committee for human research. Data safety and confidentiality was also given due consideration. The file containing identity related details was kept password protected and the filled Performa were kept in lock with key accessible only to researcher. Total 5518 consecutive births including both live born babies and still born babies were examined after taking verbal and written consent of their mothers for a visible structural anomalies to determine the overall incidence and distribution of congenital malformations. To cover all the findings of relevant history and of examination, a performa was pre-designed. According to it a complete medical history and family history for any congenital malformation, antenatal history for exposure to infection, drugs and irradiation, maternal history for age, consanguinity and parity and personal history was taken. High risk neonates were examined in detail by a neonatologist. All the babies were examined within 12 hours of birth. Thorough physical examination of

newborn babies was done. Immediate outcome of all the malformed babies was recorded during the period of mother's hospital stay and attempt was made to find out any history of congenital malformations in other family members.

Any malformed baby suspected of having syndromic congenital malformation was also confirmed by investigations e.g. ultrasonography, X-ray, echo and also by taking expert opinions of pediatrician. Data were statistically analyzed using SPSS software (trial version).

RESULTS

In the present study, we studied the total numbers of babies born in New Civil Hospital, Surat for a period of one year from 1st January 2010 to 31st December 2010. A total of 5518 babies were born out of which 75 were twins. Out of total 5518 newborns 5316 were live births and 202 were still births and out of 5316 live births 48 babies were malformed and out of 202 still births 20 babies were malformed. Total numbers of malformed babies were 68, so total point incidence of congenital anomalies turned out to be 1.23% (Table 1).

Table 1: Incidence of congenital anomalies.

| | |
|---|-------|
| Total no. of deliveries | 5443 |
| Total no. of twin deliveries | 75 |
| Total no. of new borns | 5518 |
| Total no. of malformed newborns | 68 |
| Incidence of congenital anomalies | 1.23% |
| Incidence of congenital anomalies/1000 births | 12.32 |

As far as systemic distribution of congenital malformed babies is concerned, in the present study, it was observed that out of total 5518 babies, 35 (0.63%) were having central nervous system malformations making its incidence of 6.34/1000 live births which turned out to be highest followed by gastrointestinal system (incidence of malformed babies: 5.44/1000 births), genitor urinary system (incidence of malformed babies: 1.09/1000 births), musculoskeletal system (incidence of malformed babies: 1.63/1000 births) and congenital anomalies of eyes and ears (incidence of malformed babies: 0.54/1000 births).

Commonest anomalies in central nervous system were meningomyelocele followed by hydrocephalus and anencephaly. Commonest anomalies in gastro intestinal system were bilateral cleft lip and cleft palate followed by imperforated anus and tracheo-esophageal fistula. Commonest anomalies in genito urinary system were hypospadias followed by ectopia vesicae. Commonest anomalies in musculo skeletal system were congenital talipes equino varus followed by polydactyly (Table 2).

Table 2: Distributions of newborns according to system wise incidence and distribution of congenital anomalies.

| System | Distribution of malformation | No. of malformed babies | Total no. of malformed babies (%) | Incidence/ 1000 births |
|---|--------------------------------------|-------------------------|-----------------------------------|------------------------|
| Central nervous system (including skull and spine) | Meningomyelocele | 14 | 35 (0.63) | 6.34 |
| | Hydrocephalus | 8 | | |
| | Anencephaly | 5 | | |
| | Spina bifida occulta | 4 | | |
| | Encephalocele | 2 | | |
| | Meningocele | 2 | | |
| Gastro intestinal system | Bilateral cleft lip and cleft palate | 11 | 30 (0.54) | 5.44 |
| | Imperforated anus | 10 | | |
| | Tracheo-esophageal fistula | 7 | | |
| | Cleft palate | 1 | | |
| | Omphalocele | 1 | | |
| Genito-urinary System | Hypospadias | 3 | 6 (0.11) | 1.09 |
| | Ectopia vesicae | 2 | | |
| | Absence of urethral meatus | 1 | | |
| Musculo skeletal system | Congenital Talipes equino varus | 5 | 9 (0.16) | 1.63 |
| | Polydactyly | 4 | | |
| Congenital anomalies of eyes and ears | Micro-otia | 2 | 3 (0.05) | 0.54 |
| | Anophthalmos | 1 | | |

DISCUSSION

In our study incidence of congenital anomalies was 1.23%. Other studies like Datta et al, Swain et al, Taksande A et al, Anand et al and Karla et al showed incidence of congenital anomalies were 1.24%, 1.2%, 1.91%, 2% and 1.98% respectively.^{4,6,9-11} Studies like Desai N et al and Saifullah et al showed slightly higher incidence (3.6%) than our study.^{12,13} The true incidence of congenital malformations depends upon several factors and no two studies are strictly comparable. It depends upon ethnic background, population sample (hospital or community based, live birth or total birth), nature of study (prospective or retrospective), age at the time of diagnosis, duration of follow up, autopsy rate, diagnostic facility available and enthusiasm and acuteness of physician. In the present series, low incidence in comparison to other studies is possibly because of malformations only present at birth were included. All those malformations recognized as result of autopsy study or which were diagnosed later on was excluded.

In our study, out of 202 still born babies, 20 (9.90%) babies were malformed while out of 5316 live born babies, 48 (0.90%) babies were malformed. other studies like Datta et al, Swain et al and Taksande A et al also found higher incidence of congenital malformations in still birth.^{4,6,9} Usually major malformations are

incompatible with life this may be the reason of high incidence of congenital malformation in still born babies.

In our study, out of 68 babies with congenital malformation 9 babies (13.2%) had multiple system involvement, while 59 babies (86.7%) had single system involvement. Central nervous system was the most affected system in our study. Other studies like Swain et al, Anand et al and Karla et al also found most common involvement of central nervous system while Datta et al, Taksande A et al and Desai N et al demonstrated maximum involvement of musculoskeletal system.^{4,6,9-12}

This study was conducted in a tertiary care centre with specialized maternal and neonatal care. Therefore the number of mothers and babies with complications could be more than that in the community. Hence the rate of occurrence of malformations among babies also could be more than that in the general population.

CONCLUSION

From present study we conclude that incidence of congenital anomalies of CNS was highest amongst all types of congenital anomalies (meningomyelocele being the commonest). More emphasis should be given on prevention by regular antenatal care and avoidance of known teratogens and probable teratogenic agents.

Antenatal diagnosis, genetic counseling, better diagnostic and management facilities should be provided to improve the outcome.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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