



Incidence of congenital malformations and its association with maternal risk factors: A prospective study

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Abstract

Introduction: Congenital malformations are defined as abnormalities of structures present at birth. The etiology of congenital malformations is heterogeneous. It can occur due to genetic, environmental and unknown causes. Various maternal risk factors are associated with its occurrence like advancing age, parity, socioeconomic status, previous history of malformations, maternal infections, chronic illness and nutritional diseases. Hence, this study is being conducted to see incidence of congenital malformations and its association with maternal risk factors.

Material and Methods: A prospective observational study was conducted over a period of one year at a tertiary hospital. A total of 900 women were included in the study. A detailed obstetric history was taken. Women were followed up till deliveries. Data analysis was done by SPSS for windows version 2.0.

Results: A Total of 900 deliveries were conducted, out of which there were 831 live births and 69 were stillbirths. Incidence of congenital malformations was found to be 68.89/1000 births (6.889%). Most common congenital malformation seen was central nervous system malformations (66.13%). Women with advancing age were more commonly associated with congenital malformations than younger women (highest risk in age group 25-29 years, 13.04%). Multigravida women had higher risk than primigravida. Women belonging to rural population were at higher risk than the women belonging to urban population (7.15% and 6.5% respectively). Women of lower middle class were found to have highest proportion of malformed babies (41.94%). 8.06% of women with congenital malformations had previous history of congenital malformations and 3.22% had history of previous 2 or more abortions. Mothers, who were unbooked cases, had higher incidence of congenital malformations than booked cases (7.66% and 5.94% respectively).

Conclusions: Congenital malformations pose a threat for survival of a newborn in the early neonatal period. Various maternal risk factors are associated with its occurrence. A rational and multidisciplinary approach with good communication between various specialties (obstetrician, radiologist, Pediatrician, Genetic specialist and pathologist) and parents is essential. Regular screening for high risk mother's subject should be done for early diagnosis and timely management of such cases.

Keywords: birth defects, congenital malformations, maternal risk factors

Introduction

Congenital malformations are defined as abnormalities of structures present at birth and attributable to faulty development. Congenital anomaly may be defined as all structural, biochemical and functional disorder present at birth. Thus, the term malformation must be limited to the disturbance of structures recognizable with the naked eye.

It can be a major or minor anomaly. A major anomaly is defined as one, which is severe enough to cause death or serious medical, surgical or cosmetic sequences. A minor anomaly is defined as having no surgical, medical or cosmetic importance.

According to global report on birth defects, 2006, annually 3.3 million infant's deaths are associated with birth defects, mainly major anomalies. More than 90% of infants with a serious birth defect are born in low- and middle-income countries, which lack adequate antenatal diagnostic and postnatal corrective services^[1]. In India, Various population and hospital-based studies from different parts show that 2.5% of new-born have a birth defect and congenital malformations are found to be as third most

leading cause of perinatal mortality^[2, 5].

The etiology of congenital malformations is heterogeneous and complex. Congenital malformations may be due to genetic, environmental or a combination of both these factors and unknown causes. Various maternal risk factors are also found to be associated with its occurrence like advancing age, parity, socioeconomic status, racial variation, consanguineous marriage, previous history of malformations, maternal infections, chronic illness such as diabetes, hypertension and nutritional diseases like anemia and under nutrition.

Hence, this study is being conducted to see incidence of congenital malformations and its association with maternal risk factors.

Material and Methods

A prospective observational study was conducted in department of obstetrics and Gynaecology at a tertiary hospital after clearance from institutional ethics committee (IEC), over a period of one year. A total of 900 women were included in the study. A

detailed obstetric history was taken. The diagnosis was made by USG in antenatal cases and at the time of delivery in those cases that had no previous USG reports or antenatal checkups. Women were followed up till deliveries. Data analysis was done by SPSS for windows version 2.0. The maternal risk factors included in the study are as follows:

- Maternal age at the expected date of delivery.
- Parity of woman, socioeconomic status, residential status (rural/ urban).
- History of antenatal visits and ultrasound during pregnancy (booked/unbooked case).
- Previous birth of a child with congenital malformations.
- History of two or more spontaneous abortions
- Obstetrical high-risk factors such as oligohydramnios, polyhydramnios.

Results

Total of 900 deliveries were conducted, out of which there were 831 live births and 69 were stillbirths. Total number of babies with congenital malformations were 62 out of which there were 23 live born babies, 29 still born babies and 10 were aborted (<20wks). General survey of congenital malformations seen is shown in table I.

Incidence of congenital malformations was found to be 68.89/1000 births (6.889%). Out of these 62 cases, most common congenital malformation seen was CNS malformations (66.13%) followed by musculoskeletal system malformations (24.19%) as shown in figure I.

Among maternal risk factors, it was found that women with advancing age are more commonly associated with congenital malformations than young women. The rate of fetal malformations was found to be highest in 25-29-year age group (13.04%) followed by age group >35 years (6.12%). The rate of congenital malformation was observed to be lowest in age group <19 year as shown in figure II.

It was observed that the incidence of congenital malformations was more in multigravida as compared to primigravida and maximum in second gravida (10%) followed by gravida four (8.79%). The incidence was lowest in primigravida (4.65%). The results are shown in table II.

Women belonging to rural population were at higher risk of having babies with congenital malformations than the women belonging to urban population (7.15% and 6.5% respectively) and difference was found to be statistically significant (p value <0.05) as shown in table III.

Women of lower middle class were found to have highest proportion of malformed babies (41.94%) as compared to women of upper class (1.61%). 58.06% malformed babies were belonged to combined poor & lower middle class. This high percentage indicates that congenital malformations are more in lower social class groups and it is suggested that malnutrition of pregnant mother, poor hygiene and living status could have contributed to a higher incidence in these classes.

Mothers, who were unbooked cases, were found to have higher incidence of congenital malformations as compared to mother who were booked cases, who had consumed regular iron and folic acid tablets (7.66% and 5.94% respectively) and the difference is statistically significant (p <0.05). The results are shown in table IV.

In the present study, an association was found between various obstetrical conditions and the occurrence of congenital malformations (shown in table V). 9 women (14.52%) were found to have oligohydramnios. Most common congenital malformations seen in these were renal malformations (4 cases) and lower limb deformity (4 cases). Polyhydramnios was an offending risk factor in 8 cases (12.92%) of congenital malformations, of which most common anomaly was CNS defects (6 cases of neural tube defects and 5 cases of anencephaly).

8.06% of women with congenital malformations had previous history of congenital malformations and 3.22% had history of previous 2 or more abortions. One woman had history of diabetes mellitus, who delivered a baby with congenital heart disease.

Discussion

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. It is defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.

Congenital anomalies are important causes of infant and childhood deaths, chronic illness and disability. It can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies.

An estimated 303 000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies according to WHO fact sheet 2016^[6]. Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known genetic, environmental and other causes or risk factors. Thereby, we are going to discuss about incidence and association of congenital malformation with various maternal risk factors.

The present study suggest that overall incidence of congenital malformations is 68.89/ 1000 births (6.889%). The CNS malformations are the most common among them (66.13 %) followed by malformations of musculoskeletal system (24.19%). However, in various studies incidence found is comparatively lower (0.3 to 3.6%)^[7, 10].

In 2018, a study was conducted by Bhide *et al.*^[11], to derive an estimate of the birth prevalence of congenital anomalies in India. The search was carried out in PubMed. The pooled prevalence of congenital anomaly affected births was found to be 184.48 per 10,000 births (18.84/1000 live births) among 802,658 births. Anomalies of the musculoskeletal system were highest among live births while the prevalence of central nervous system defects was highest among stillbirths. Anencephaly and talipes were the most commonly reported anomalies.

This wide variation in the incidence of malformations between different studies can be due to various factors such as population sampled, geographical and racial difference, selection of cases-live born, stillbirth or both and age of the baby when diagnosis is made. It may also be due to the fact that this study is conducted at a referral institute and more cases with congenital malformations were referred from periphery.

A positive association was found between incidence of congenital malformations and advancing maternal age. In the present study, maximum incidence of congenital malformations was observed in age group 25 – 29 yr. Similar results were seen

in study by Kulshrestha *et al.* [12] and Padmanabhana *et al.* [13]. However, Khanum S *et al.* [14] found higher incidence in age group >35 yr.

In 2019, a study was published by R Padmanabhan *et al.*, to determine the prevalence and pattern of CMs among neonates in a teaching hospital. In 2132 babies, malformations were seen in 87 babies (4.08%). It was found that the incidence of congenital malformations was highest in age group 21-30 years (87.4%) followed by women of age >31years (9.2%).

This positive association can be attributed to the fact the following factors:

- Increasing age leads to higher chances of chronic illness, thus decreasing the immunity.
- Also, with advancing age, risk of genetic mutation increases. Maternal age increases the risk of chromosomal abnormalities, including down syndrome [7].

In the present study, it was found that congenital malformations are more common in multigravida women than primigravida. Results were in accordance with study by Khanum S *et al.* [14] and Malik M *et al.* [15], in which all of them found higher incidence in multipara.

A study was done by Malik M *et al.* in 2019, to find association between maternal risk factors and congenital anomalies of infants. A community-based retrospective and cross-sectional study was carried out in 23 rural sub-centres of block Beri, district Jhajjar (Haryana, India) among 920 mothers. The prevalence of congenital malformations was 1.2%. Most common congenital malformations were cleft lip/palate (18.18%) and hydrocephalus (18.18%). In this study, it was found that risk of congenital malformations is more in multiparas (3.3%) than the primigravida (1.8%).

In the present study, women belonging to rural population were at higher risk of having babies with congenital malformations than the women belonging to urban population. The results are comparable to study done by Jain VK *et al.* [16] who observed significant difference between rural and urban population.

According to report of WHO, the Expert Committee 2016 [7], It is estimated that about 94% of severe congenital anomalies occur in low- and middle-income countries. Similar results were obtained by various studies [17, 19] as well as in the present study. This could be because of poor nutrition, poor hygiene, thereby increasing the exposure to agents or factors such as infection and alcohol, poorer access to healthcare and screening.

Mothers, who were unbooked cases, were found to have higher incidence of congenital malformations as compared to mother who were booked cases (7.66% and 5.94% respectively) and the difference is statistically significant (p <0.05).

Similar results were found by study of Marwah *et al.* [20] done in 2018. A total of 1554 deliveries were included, of which 69 babies had congenital malformations (4.44%). It was found that women belonging to rural area had higher incidence of congenital malformations as compared to women of urban area (5.10% and 1.20% respectively). 66 out of the total 69 mothers with malformed babies dwelled from rural areas of the region. Also, a higher incidence of congenital malformations was observed in women who were unbooked cases than those booked for antenatal care (5.45% and 3.18% respectively). The incidence of congenital malformations was significantly higher in the lower

strata (6.26%) and it decreased as the socioeconomic status of women improved.

In the present study, polyhydramnios was present in 12.9% cases which are comparable to study done by Kalra *et al.* [21] who found 16.6 % cases with polyhydramnios. In our study oligohydramnios was associated with 14.52% cases which is much high as compared to the study done by Martinez Frias *et al.* [22] (3.7%). In this study by Martinez Frias *et al.* [22], a series of 27,145 congenitally malformed babies were included. Among these, polyhydramnios was seen in 927 cases (3.69%) while oligohydramnios was found in 755 cases (3.01%). Most common congenital malformations seen in cases with polyhydramnios was cardiovascular defects (16.69%) followed by lung defects (8.91%). In oligohydramnios also, most common anomaly seen was cardiovascular defects (10.17%) followed by hydrocephalus (5.93%).

Overall recurrence rate of CNS malformations in present pregnancy is 8.06% and recurrence of NTD's is 4.83%. This is comparable to studies done by Verma *et al.* [23] and Hansmann M *et al.* [24], who found recurrence of NTD's between 3to 5%.

Table 1: General Survey Of Congenital Malformations

	Total no of cases	Cases with Congenital malformations	Percentage (%)
Total no of deliveries	900	62	68.81%
Live born babies	831	23	2.76%
Still births	69	29	42.02%
Male babies	526	30	5.70%
Female babies	372	30	8.06%

Table 2: Incidence of congenital malformations in rural/urban population

Residential status	Babies with congenital malformations	Normal babies	Total babies	Incidence of malformations/ 1000 births
Urban	24 (38.71%)	345	369	65.04 (6.5%)
Rural	38 (61.29%)	493	531	71.56 (7.15%)
Total	62	838	900	

Table 3: Incidence (%) Of Congenital Malformations In Relation To Gravidity

Gravidity	Cases with congenital malformations	Cases without congenital malformations	Total cases	Percentage (%) of cases with congenital malformations
G1	19	389	408	4.65%
G2	23	207	230	10%
G3	12	159	171	7.01%
G4	8	83	91	8.079%

Table 4: Incidence of congenital malformations in booked and unbooked cases

Status of patient	Babies with malformations.	Normal babies	Total Babies	Incidence of malformations/ 1000
Booked	24 (38.71%)	380	404	59.41 (5.94%)
Unbooked	38 (61.29%)	458	496	76.61 (7.66%)
Total	62	838	900	

Table 5: Association Between Obstetrical Conditions And Congenital Malformations

High risk factor	No. of cases	Percentage (%)
Oligohydramnios	9	14.52
Polyhydramnios	8	12.90
Prev. history of congenital malformations	5	8.06
Prev. H/o 2 or more abortions	2	3.22%
Diabetes Mellitus	1	1.61%

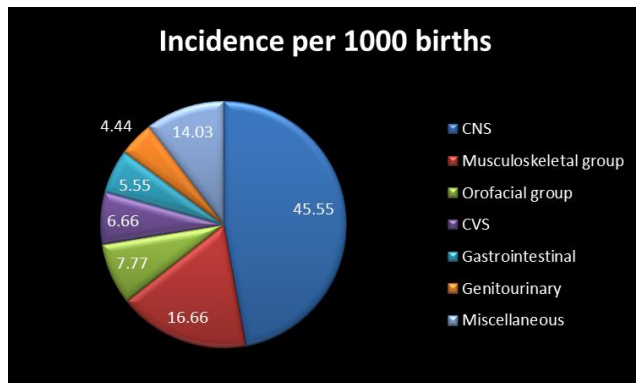


Fig 1: Pie Chart Showing Distribution Of Congenital Malformations In Different Systems In Terms Of Incidence Per 1000 Births.

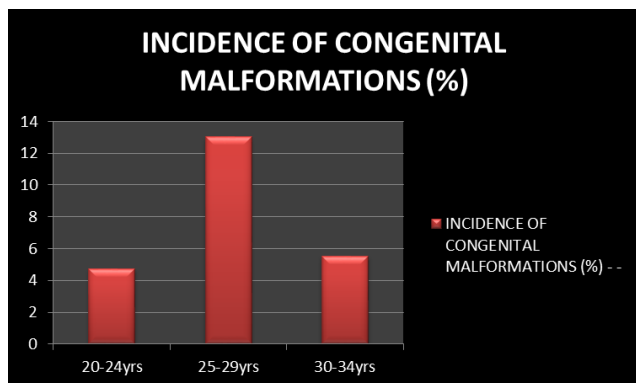


Fig 2: a bar graph showing association between maternal age and incidence of congenital malformations.

Conclusions

Congenital malformations pose a threat for survival of a newborn in the early neonatal period. Prevention of congenital malformation can be achieved by proper marriage counseling for avoiding early and consanguineous marriages, periconceptional folate consumption, regular antenatal checkup, screening for maternal high-risk factors and need for at least one antenatal ultrasound scan at around 18-20 weeks to rule out gross congenital malformations. A rational and multidisciplinary approach with good communication between various specialties (obstetrician, radiologist, Pediatrician, Genetic specialist and pathologist) and parents is essential.

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