

## Study of incidence and outcome of structural malformed foetus in delivered patients

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### Abstract

**Introduction:** Congenital fetal anomalies are one of the most threatening complications which are prevalent in the society associated with severe morbidity and mortality in the newborn foetus. Congenital anomalies account for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India. The etiology of congenital abnormality may be genetic or environmental.

**Aim and Objective:** The present study was carried out with the aim to determine the overall incidence and outcome of congenital structural malformations in delivered patients, to identify the most common structural anomaly and study the socio-demographic correlates with structural anomalies and compared them to previous studies.

**Materials and Methods:** The study was conducted on all obstetrics patients with consent who have delivered with fetal congenital malformations in Medical College Pune, Maharashtra over a period of 2 year satisfying inclusion and exclusion criteria. The questionnaire was prepared included the information regarding mothers age, gender birthweight, religion and socioeconomic, past and personal history was included. Data management and analysis was done using Epi-info software. The frequency distribution and graph was prepared. The categorical variables was assessed using Pearson chi-square.

**Results:** It was observed that the overall incidence of structural malformed foetus delivered came out to be 1.2%. Low Birth weight, Low socio economic status and female gender of child are commonly involved in congenital structural malformations. There was no significant association observed between system involved with gender of child, gestational age or history of abortion. The most common anomaly found is related to GIT followed by the Genitourinary.

**Conclusion:** We conclude that gastrointestinal, genitor-urinary, cardiovascular and nervous systems are commonly involved in congenital structural malformations. Among the gastro-intestinal system congenital hypertrophic pyloric stenosis and Anorectal malformations are the commonest ones. However Low Birth weight, Lower socio economic status and female gender of child are commonly involved in congenital structural malformations.

**Keywords:** Congenital, Abnormalities, Structural malformations, New born, Low birth weight.

### Introduction

Congenital fetal anomalies are one of the most threatening complications which are prevalent in the society associated with severe morbidity and mortality in the new born fetus or neonates.

According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.<sup>1-3</sup>

Congenital malformations are single or multiple defects of the morphogenesis of organs identifiable at birth or at the intrauterine life. There is an intrinsically abnormal developmental process, with the early development of a tissue or organ being arrested, delayed, or misdirected. These anomalies can be caused by a teratogen or abnormalities in a gene, a group of genes, or a chromosome.<sup>4</sup>

Congenital malformations contribute highly to prenatal mortality and postnatal physical defect. Aggregates of literature indicates that congenital malformations are present in one of every three babies that die in the world and their effects on a child vary depending greatly on the severity of the defect and whether or not other medical problems are present. 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.<sup>5</sup>

Congenital anomalies account for 8% to 15%<sup>6,7</sup> of perinatal deaths and 13% to 16% of neonatal deaths in India.<sup>8,9</sup> The etiology of congenital abnormality may be genetic (30–40%) or environmental (5–10%). Among genetic causes, chromosomal abnormality makes up about 6%, single gene disorders about 25%, and multifactorial factors 20–30%. In about 50% of cases, the cause is not known.<sup>10</sup>

Congenital abnormality plays a major role in morbidity and mortality of children especially at the perinatal period. The treatment and rehabilitation of these children with congenital abnormality is very costly, hence the need to identify causative and risk factors and prevent them early,<sup>4</sup> where possible. Early recognition of anomalies is also important for planning and care. Parents are likely to feel anxious and guilt on learning of the existence of a congenital anomaly and this require sensitive counselling.<sup>2</sup>

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. The present study was carried out with the aim to determine the overall rate of congenital malformations, incidence in live births and stillbirths, as well as incidence affecting various organ

systems, at a medical college hospital in Maharashtra and compare them to previous studies.

### Aims and Objectives

The present study was carried out with the aim to determine the overall incidence and outcome of structural malformed foetus in delivered newborns, To find most common structural anomaly in newborn cases further to study the socio-demographic correlates of newborns with structural anomaly.

### Materials and Methods

Study was conducted on all obstetric patients who have delivered with fetal congenital malformation on over a period of 2 years from July 2016 to July 2018. Institutional ethical committee clearance was obtained with inclusion and exclusion criteria as

#### Inclusion Criteria

1. All obstetric patients who have delivered with fetal congenital malformation.
2. Those who have given informed consent.

#### Exclusion Criteria

1. Medicolegal cases.
2. Women who did not given consent

Written informed consent was taken from all the selected cases with detailed history taking and examination was done. A predesigned semi-structured questionnaire was prepared. The questionnaire included the information regarding mothers age, gender, birth weight, religion and socioeconomic status. It also included information regarding parity, birth order, gestational age, history of abortion and still births, family history of congenital disease, consanguinity.

Data management and analysis was done using Microsoft excel and Epi-info software. The frequency distribution and graph was prepared for the variables. The categorical variables was assessed using Pearson chi-square. The test was considered significantly only if the p value comes out to be less than 0.05.

### Results

In our study total 52 number of congenital malformed cases are included which met our study criteria and on the basis of system involvement i.e. the distribution of cases according to System involvement in congenital disease, it was observed 9 (17.3%) cases had cardiovascular involvement, 5 (9.6%) cases had Cleft lip and palate, 11 (21.2%) cases had digestive involvement, 10 (19.2%) cases had genito-urinary involvement, 7 (13.5%) cases had musculoskeletal involvement, 9 (17.3%) cases had nervous involvement and 1 (1.9%) cases had respiratory involvement (Table 1).

On the basis of type of congenital anomaly most common found to be gastro-intestinal system in which

Congenital hypertrophic pyloric stenosis 3 (27.2%) and Anorectal malformations 3 (27.2%), among the genitor-urinary system Hypospadias 4 (40%) and Undescended testicle 3 (30%), among the cardiovascular system Ventricular Septal defect 4 (44.4%) and Atrial Septal defect 3 (33.3%) and among the nervous system Hydrocephalus 3 (33.3%), Meningocele 2 (22.2%) and Encephalocele 2 (22.2%) are commonly seen (Table 2).

In present we correlated different socioeconomic factors with the different congenital malformation to find out the significant correlation. Present study includes 52 total cases out of which 25 (48.1%) children were male and 27 (51.9%) children were female and its correlation with different congenital malformation is not significant with P value 0.47 (Table 3). According to birth weight it was found that 10 (19.2%) have birth-weight less than 2000 grams, 25 (48.0%) children have birth-weight between 2000-2499 grams and its correlation with different congenital malformation is not significant (Table 4). The mean birth weight of the children was  $2348 \pm 333.7$  grams. According to socio economic status 9 cases (317.3%) belongs to high socio-economic group, 7 (13.4%) belongs to middle socio-economic group and 36 (69.2%) belongs to lower socio-economic group. On basis of gestational age it was observed that 18 (34.6%) cases were preterm and 34 (65.4%) cases were of term gestational age and its correlation with different congenital malformation is not significant with P value 0.32 (Table 5). History of abortion showed that in 11 cases (21.2%) abortion history was present and 41 (78.8%) cases abortion history was absent and its correlation with different congenital malformation is not significant with P value 0.19 (Table 6). Antenatal visit during pregnancy is important for early screening and prevention of malformed fetus, in our study it was observed that 17 (32.7%) cases had nil ANC visit, further 10 (19.2%) cases had single ANC visit, 15 (28.8%) cases had 2 ANC visit, 5 (9.6%) cases had 3 ANC visit, 4 (7.7%) cases had 4 ANC visit and 1 (1.9%) cases had 5 ANC visit. Iron & Folic acid tablet consumption history was significant and observed that 17 (32.7%) cases had consumed all given IFA tablets and 34 (65.4%) cases had not consumed IFA tablets.

**Table 1: Distribution of cases according to System involvement**

System	Frequency	Percent
Cardiovascular	9	17.3
Cleft lip and palate	5	9.6
Digestive	11	21.2
Genitourinary	10	19.2
Musculoskeletal	7	13.5
Nervous	9	17.3
Respiratory	1	1.9
Total	52	100.0

**Table 2: Distribution of cases according to type of congenital anomaly**

System	Type of congenital anomaly	No.
Respiratory	Choanal atresia	1
Cardiovascular	VSD	4
	ASD	3
	Tetralogy of fallot	2
Nervous	Hydrocephalus	3
	Meningocele	2
	Encephalocele	2
	Meningomyelocele	1
	Spina bifida	1
Cleft lip and palate	Cleft lip and palate	5
Digestive	Hirschsprungs disease	2
	Anorectal malformations	3
	Congenital absence, stenosis of anus	1
	Congenital hypertrophic pyloric stenosis	3
	Meconium Ileus	1
	Congenital absence, atresia and stenosis of small intestine	1
Genitourinary	Hypospadias	4
	Undescended testicle	3
	Congenital hydrocele	1
	Cloacal anomaly	1
	Congenital hydronephrosis	1
Musculoskeletal	Gastroschisis	2
	Polydactyly	2
	Congenital umbilical hernia	3

**Table 3: Comparison of system involvement with gender**

System involved	Male	Female	Total
Cardiovascular	4	5	9
Cleft lip and palate	1	4	5
Digestive	5	6	11
Genitourinary	6	4	10
Musculoskeletal	3	4	7
Nervous	5	4	9
Respiratory	1	0	1
Total	25	27	52

**Table 4: Comparison of system involvement with birth weight**

System involved	Birth Weight (in grams)				Total
	1500-1999	2000-2499	2500-2999	3000-3499	
Cardiovascular	1	6	1	0	6
Cleft lip and palate	1	1	3	0	5
Digestive	3	6	2	0	11
Genitourinary	1	7	2	0	10
Musculoskeletal	2	2	2	1	7
Nervous	1	3	5	0	9
Respiratory	0	0	1	0	1
Total	10	25	19	1	52

**Table 5: Comparison of system involvement with gestational age**

System involvement	Gestational age		Total
	Preterm	Term	
Cardiovascular	4	5	9
Cleft lip and palate	1	4	5
Digestive	6	5	11
Genitourinary	3	7	10
Musculoskeletal	2	5	7
Nervous	2	7	9
Respiratory	0	1	1
Total	18	34	52

**Table 6: Comparison of system involvement with history of abortion**

System involvement	H/O Abortion		Total
	Present	Absent	
Cardiovascular	2	7	9
Cleft lip and palate	0	5	5
Digestive	3	8	11
Genitourinary	1	9	10
Musculoskeletal	3	4	7
Nervous	2	7	9
Respiratory	0	1	1
Total	11	41	52

## Discussion

The present study is aimed to identify the most common congenital malformed anomaly in delivered patients and its incidence and it was found that most common anomaly found is related to gastro intestinal system followed by genito-urinary system, other system commonly involved are cardiovascular, nervous, respiratory and musculoskeletal systems, another similar study conducted by Chowdhury Fazle Mubarak Bari et al,<sup>11</sup> demonstrates 26 (9.7) cases had cardiovascular involvement, 19 (7.1) cases had chromosomal abnormalities, 18 (6.7) cases had Cleft lip and palate, 72 (27.0) cases had digestive involvement, 42 (15.7) cases had genito-urinary involvement, 40 (15.0) cases had musculoskeletal involvement, 42 (15.7) cases had nervous involvement and 3 (1.1) cases had respiratory involvement. Similarly in the study conducted by Wills V et al,<sup>12</sup> it was observed 31 (20.5) cases had cardiovascular involvement, 4 (2.6) cases had multisystem involvement, 14 (9.3) cases had digestive involvement, 43 (28.5) cases had genito-urinary involvement, 18 (11.9) cases had musculoskeletal involvement, 18 (11.9) cases had nervous involvement and 13 (8.6) cases had respiratory involvement. In one another study conducted by K Fatema et al<sup>13</sup> it was observed that 6.68% cases had digestive involvement, 23.33% cases had genito-urinary involvement, 5% cases had musculoskeletal involvement, 46.67% cases had nervous involvement and 11.67% cases had multiple congenital anomalies and 5% had non immune hydrops fetalis.

On the basis of gender in the present study, it was observed that 25 (48.1%) children were male and 27 (51.9) children were female. In the study conducted by Obu et al<sup>2</sup> it was observed that 8 (47.1) children were male and 9

(52.9) children were female. The results of Obu et al are comparable to the findings of our study where female cases are more than the male cases. It was observed that there are no major differences among both the genders in system involvement with congenital anomaly. In another study by Chowdhury Fazle Mubarak Bari et al<sup>11</sup> it was observed that 141 (52.8) children were male, 124 (46.4) children were female and 2 (0.7) children were having indeterminate sex. Similarly study conducted by K Fatema et al<sup>13</sup> it was observed that 41 (68.3) children were male and 19 (31.7) children were female.

According to birth weight of the child the study conducted by Obu et al<sup>2</sup> it was observed 5 (29.4) children have birth-weight less than 2500 grams, 6 (35.3) children have birth-weight between 2500-4000 grams and 1 (5.9) child has birth-weight more than 4000 grams. The mean birth weight of the children was 30600 ± 480 grams. In another study conducted by K Fatema et al<sup>13</sup> it was observed that it was observed 32 (53.3) children have birth-weight less than 2500 grams and 28 (46.7) children have birth-weight more than 2500 grams.

In view of gestational age, in the present study, it was observed that in term children nervous system 7 (20.5) cases and genitourinary system 7 (20.5) cases was commonly involved while in preterm children, digestive system 6 (33.3) cases and cardiovascular system 4 (22.2) cases was commonly involved. There was no significant difference between gestational age and type of system involved in congenital malformations with p value more than 0.05. Similarly In the study conducted by Wills V et al,<sup>12</sup> it was observed that 42 (27.8) cases were preterm and 109 (72.2) cases were term gestational age. The findings of Wills V study are comparable to present study. In the study

conducted by Amar Taksande et al,<sup>3</sup> it was observed that 20 (11.2) cases were preterm and 156 (87.1) cases were term gestational age.

On basis of history of abortion in the present study, it was observed that among those who have positive of abortion, 2 have cardiovascular malformations, 3 have digestive system involvement and musculoskeletal involvement, 2 have nervous system involvement. Few other studies conducted by Chowdhury Fazle Mubarak Bari et al,<sup>11</sup> it was observed that in 23 (8.6) abortion history was present and 244 (91.4) cases abortion history was absent. In another the study conducted by K Fatema et al<sup>13</sup> it was observed that in 26 (43.3) abortion history was present and 34 (56.7) cases abortion history was absent. In the study conducted by Wills V et al,<sup>12</sup> it was observed that in 19 (12.7) abortion history was present and 132 (87.3) cases abortion history was absent.

In our study the correlation with ante natal visit showed that 17 (32.7) cases had nil ANC visit, 10 (19.2) cases had single ANC visit, 15 (28.8) cases had 2 ANC visit, 5 (9.6) cases had 3 ANC visit, 4 (7.7) cases had 4 ANC visit and 1 (1.9) cases had 5 ANC visit. Similarly in the study conducted by Chowdhury Fazle Mubarak Bari et al,<sup>11</sup> it was observed that 112 (41.9) cases had nil ANC visit, 115 (43.1) cases had irregular ANC visit and 40 (15.0) cases had regular ANC visit.

On the basis of Iron and folic acid consumption present study shows that 17 (32.7) cases had consumed all given IFA tablets and 34 (65.4) cases had not consumed IFA tablets. In the study conducted by Chowdhury Fazle Mubarak Bari et al,<sup>11</sup> it was observed that 82 (30.7) cases had consumed all given IFA tablets and 185 (69.3) cases had not consumed IFA tablets.

## Conclusion

On the basis of our study we conclude that the overall incidence of structural malformed foetus delivered came out to be 1.2%. Gastrointestinal, genito-urinary, cardiovascular and nervous systems are commonly involved in congenital structural malformations. Among Gastro-intestinal system, congenital hypertrophic pyloric stenosis and Anorectal malformations were commonly found. In Genitor-urinary system, Hypospadias and Undescended testicle was common. In Cardiovascular system, Ventricular Septal defect and Atrial Septal defect was found to be more common. Among the nervous system, Hydrocephalus, Meningocele and Encephalocele are commonly seen.

Overall most common anomaly found in our study is related to Gastro-intestinal in which Anorectal malformations and congenital hypertrophic pyloric stenosis found to have equal prevalence. Although Birth weight less than 2500 grams, Low socio economic status and female gender of child are commonly involved in congenital structural malformations in our study. However there was no significant association observed between system involved and gender of child, gestational age and history of abortion.

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