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**Original Research Article**

# Profile of congenital malformations in a tertiary care level neonatal intensive care unit

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## ABSTRACT

**Background:** Congenital malformations are important causes of infant and childhood deaths, chronic illness and disability.
**Methods:** This prospective observational study was conducted over a period of one year in Princess Esra Hospital, a tertiary level neonatal intensive care unit, Deccan College of Medical Sciences, Hyderabad, Telangana, India. All new-borns admitted to the neonatal intensive care unit over a period of 1 year from June 2014 to June 2015 were included in the study.
**Results:** The incidence of congenital malformations (CMF) in the present study is 3%. Among the major congenital malformations, the central nervous system (CNS) and gastrointestinal (GI) malformations were more frequent than others contributing to 28.9% each. The male neonates were more frequently involved than females. Association of low birth and prematurity with increased risk of congenital malformations is also documented.
**Conclusions:** Early prenatal diagnosis and management of common anomalies is strongly recommended.

**Keywords:** CMF, CNS, GI, Prematurity

## INTRODUCTION

Congenital malformations (CMF) are structural defects resulting from prenatal insults during the period of embryogenesis. They are an important cause of neonatal mortality both in developed and developing countries. Congenital anomalies are important causes of infant and childhood deaths, chronic illness and disability. Neonates with congenital malformation shave wide differences in physiology, anatomy, diseases, immunity and response to stress as compared to older patients.

Outcome depends on the availability of antenatal diagnosis, improved surgical skills and technologies, sophisticated neonatal intensive care unit, availability of total parenteral nutrition and adequate staff.

In developing countries however, there is delay in presentation and lack of medical facilities making congenital malformations to be associated with unacceptably high morbidity and mortality.

The reported incidence varies from 3% for major congenital malformations and 12-15% for minor defects. The major CMF contribute to 15% of perinatal mortality and 13-16% of neonatal deaths in India.[1,2]

The aim of this study is to provide information on the clinical profile of congenital malformations in a tertiary care level neonatal intensive care unit and highlight the importance of early diagnosis and surgical correction of malformed babies which offer best chances of survival.

## METHODS

This prospective observational study was conducted over a period of one year in Princess Esra Hospital, a tertiary level neonatal intensive care unit, Deccan College of Medical Sciences, Hyderabad, Telangana, India. All new-borns admitted to the neonatal intensive care unit over a period of 1 year from June 2014 to June 2015 were included in the study. The data was collected prospectively. The diagnosis of congenital anomalies was based on clinical examination and appropriate investigations like radiography, ultrasonography, chromosomal analysis etc. Surgical cases are handled by pediatric surgeons and pediatrician. Case notes and admission records in newborn unit were used to extract the following information: Sex, age at presentation, date of admission, maturity, surgical condition, treatment and prognosis (outcome of the management). A database was created recording the gestation, birth weight, type of malformation, diagnosis, and outcome.

The neonates were then classified into major and minor congenital malformation groups and also into syndromic and non-syndromic variants. The major malformations were further categorized into central nervous system, cardiovascular gastrointestinal, genitourinary and miscellaneous malformations. Outcome of the babies were classified into three groups as discharged after treatment, died during the course of treatment and left against medical advice (LAMA). Standard statistical method was used to define the measure of outcome.

## RESULTS

During the four-year study period in neonatal ICU, there were 4120 neonates admitted and 123 neonates presented with malformations. Thus, the incidence of CMF in the present study is 3 percent. Among these, 22 (18%) were inborn, the rest were brought directly by parents or referred from the neighboring units. The mean age of admission was 8 days with a range of 1-28 days. The male to female ratio was 2:1 (82 males, 41 females). 37 (30%) neonates were premature. The mean birth weight of babies was 2100 gm (900g-3700g). 71 (57.7%) neonates had minor CMF while the rest 52 (42.3%) had major CMF.

**Table 1: Distribution of CMF as major and minor categories.**

| CMF | Number | % |
|---|---|---|
| Minor | 71 | 57.7 |
| Major | 52 | 42.3 |
| Total | 123 | |

Among the major CMF the CNS and GIT malformations were more frequent than others contributing to 28.9% each (15 neonates). Totally 15 neonates (28.9%) had CNS malformations which included congenital hydrocephalus, meningocoele and meningomyelocoele and encephalocele. Mean age of presentation was 1.4 days (range 2–4 days). 1 neonate had Arnold Chiari malformation and 2 neonates had dandy walker syndrome. Meningocoele and meningomyelocoele was seen in 8 neonates (15%) out of which 5 neonates (62.5%) had Lumbosacral meningomyelocoele followed by thoracic (12.5%), cervical (12.5%) and sacral (12.5%). Lipomeningomyelocoele accounts for 12.5% cases of lumbosacral meningomyelocoele and none had more than one meningomyelocoele. VACTERAL association was seen in 1 neonate (25%) with paraparesis.

**Table 2: Distribution of minor CMF with and without syndromic association.**

| Minor CMF | Number | % |
|---|---|---|
| Minor with syndrome | 10 | 9.9 |
| Minor without syndrome | 91 | 90 |
| Total | 101 | 99.9 |

The GIT malformations were seen in 15 neonates. Small or large bowel atresia was seen in 5 neonates with 3 cases of duodenal atresia, 1 case of jejuna atresia and 1 case of ileal atresia. Mean age of presentation for intestinal atresia was 3 days (range 1-7 days). Clinical manifestations included bilious emesis, aspirates in neonates, upper abdominal distension and feeding intolerance. Diagnosis was achieved in most instances by plain abdominal radiographs which demonstrated the characteristic “double-bubble” sign.

**Table 3: Distribution of major CMF with and without syndromic association.**

| Major CMF | Number | % |
|---|---|---|
| Major CMF with syndrome | 15 | 28.9 |
| Major CMF without syndrome | 37 | 71 |
| Total | 52 | |

Upper gastrointestinal contrast enhanced examination showed partial duodenal obstruction in 2 neonates with duodenal stenosis. Down’s syndrome was seen in 2 cases of duodenal atresia and 1 case of jejuna atresia. Associated anomalies were present like cardiac anomalies, renal anomalies and skeletal abnormalities.4 neonates had Hirschsprung’s Disease (HD) during the study period.

**Table 4: System wise classification of major CMF.**

| Type of CMF | Number | % |
|---|---|---|
| CNS | 15 | 28.9 |
| Respiratory | 10 | 19.1 |
| GIT | 15 | 28.9 |
| GUT | 12 | 23.1 |
| Total | 52 | 100 |

3 had classical short segment, 1 had ultra-short segment, none had long segment or total colonic or severe type.

**Table 5: Profile of major CMF.**

| System | Type of CMF | Number |
|---|---|---|
| CNS | Congenital hydrocephalus | 5 |
| | MMC | 8 |
| | Encephalocele | 2 |
| Respiratory | CDH | 3 |
| | CLE | 1 |
| | CCAM | 1 |
| | TEF | 5 |
| GIT | Duodenal atresia | 3 |
| | Jejunal atresia | 1 |
| | Ileal atresia | 1 |
| | malrotation | 1 |
| | IHPS | 1 |
| | Hirschsprung's | 4 |
| | anorectal | 2 |
| | gastroschisis | 1 |
| | examphalocele | 1 |
| Gut | PUV | 3 |
| | PUJ | 9 |
| Total | 52 | |

The male to female ratio was 2:1. 1 neonate had malrotation of gut with or without midgut volvulus Mean age of presentation was 14 days. Most common clinical features were bilious vomiting and failure to pass meconium. In this case malrotation was associated with midgut volvulus. 1 neonates had Infantile Hypertrophic Pyloric Stenosis (IHPS) who presented with non-bilious projectile vomiting, gross dehydration and non-acceptance of food. Pyloric tumour was palpable and visible gastric peristalsis was evident. Mainstay of diagnosis was clinical and confirmed by ultrasonography finding. Abdominal wall defects were seen in 2 neonates with 1 each case of exomphalos and gastroschisis. Mean age of presentation was 3 days (range 1-6 days). Anorectal malformations were seen in 2 neonates and the mean age of presentation was 6.2 days (range 1-30 days).

**Table 6: Syndromic associations of major CMF.**

| Type of syndrome | Number | % |
|---|---|---|
| Downs (Trisomy 21) | 3 | 5.7 |
| Turners | 1 | 1.9 |
| Edwards (Trisomy 18) | 3 | 5.7 |
| Patau's (Trisomy 13) | 1 | 1.9 |
| Criduchat | 1 | 1.9 |
| VACTERAL association | 2 | 3.8 |
| CHARGE association | 1 | 1.9 |
| Arnold chiari malformation | 1 | 1.9 |
| Dandy walker syndrome | 2 | 3.8 |
| Total | 15/52 | 28.8 |

10 neonates had respiratory malformations. Oesophageal atresia or tracheo oesophageal fistula was noted in 5 neonates. 1 had pure oesophageal atresia associated with downs syndrome and the most common type was type III-B which was seen in 4 neonates. 2 neonates were associated with VACTERAL anomalies. Most common associated anomaly was cardiac anomalies followed by gastrointestinal and musculoskeletal anomalies. Congenital diaphragmatic hernia (CDH) was seen in 3 neonates, the mean age of presentation was 2 days (range 1–3 days). Male to female ratio was 2:1, left sided in 2 cases and on right side in 1 case.

**Table: 7: Clinical manifestations of major CMF.**

| Clinical features | Number | % |
|---|---|---|
| Large head | 5 | 9.6 |
| Respiratory distress | 10 | 19.2 |
| Evidence of PPHN | 8 | 15.3 |
| Cyanosis | 10 | 19.2 |
| Frothing of saliva | 5 | 9.6 |
| Abdominal distention | 12 | 23 |
| Bilious vomiting | 7 | 13.4 |
| Non-bilious vomiting | 3 | 5.76 |
| Delayed/not passing meconium | 6 | 11.5 |
| Visible swelling/sinus | 10 | 19.2 |

Most common presentation was respiratory distress followed by cyanosis. 2 cases were antenatally diagnosed. During the 4 years study period, only 1each case of congenital lobar emphysema (CLE) and Congenital cystic adenomatoid malformation (CCAM) had been admitted with respiratory distress, and they were antenatally diagnosed.

Among genito-urinary malformations posterior urethral valve (PUV) was seen in 3 cases. They presented on 10th day of life with difficulty and crying during micturition, dribbling of urine and palpable and enlarged kidney. Presence of hypertrophied and distended bladder on bimanual palpation was seen. Neonate had raised blood urea and serum creatinine levels. The mainstay of diagnosis was clinical, supported by KUB, ultrasound and voiding cystourethrogram. Endoscopic valve ablation was the primary mode of treatment. There were 9 cases of pelvi uretoro junction obstruction (PUJO) admitted to our unit during this 4-year period. The mean age of presentation was 2 days (1-4days) and 6 cases (66.67 %) were diagnosed antenatally. The male to female ratio was 2:1 and common manifestation was lump abdomen, 67% were on right side, 22% were on left side and 11% were bilateral. Post-natal ultrasound confirmed the findings. All neonates received uro prophylaxis with amoxicillin.

## DISCUSSION

Congenital anomalies or birth defects can be defined as structural or functional anomalies including metabolic disorders that occur during intrauterine life and can be identified prenatally, at birth or later in life.[3] An estimated 303000 newborns die within 4 weeks of birth every year worldwide due to congenital anomalies. With improved control of infections and nutritional deficiency diseases, congenital malformations have become

important causes of perinatal mortality in developing countries like India.[4]

In the present study the prevalence of congenital malformations in newborns were 3% which is comparable with the earlier studies from India which reported incidence of 2.72% and 1.9%.[5,6] Reports also exist from different parts of the world showing different frequency of congenital malformations.[7,8] Other researchers have also observed nearly similar results in the prevelance of congenital malformations.[5-9] Kalra et al reported that the CNS defects have highest incidence whereas Suguna bai et al reported higher incidence of gastrointestinal malformations.[10,11] Mathur et al reported that the musculoskeletal abnormalities were the commonest.[12] In the present study, both CNS and GI systems showed increased frequency followed by respiratory malformations. This was comparable with the studies conducted by others.[12-17] The incidence of malformations was more in male babies compared to females[4,5] and in low birth weight neonates.[4] Present study also documented increased association of congenital malformations with prematurity.[12] In a study by Shija JK in Tanzania 3% of NICU admissions were due to congenital malformations.[19] The incidence of birth defects is higher in the hospitals where autopsies are performed. Some centres report a higher incidence due to more autopsy rates.[4,18] Congenital malformations are one of the important causes of infant mortality and morbidity. They can be reduced by regular antenatal visits and prenatal diagnosis. We found neurological defects as commonest malformation which can be easily prevented by preconceptional folic acid. High incidence of neural tube defects indicates maternal malnutrition which needs significant attention and management.

## CONCLUSION

Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination when needed. Early diagnosis and surgical correction of the malformed babies offer the best chance of survival.

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