

## Original Research Article

# A study of gross congenital malformation at birth

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### ABSTRACT

**Background:** A congenital anomaly is a structural anomaly of any type that is present at birth. Congenital anomalies may be induced by genetic or environmental factors. Most congenital anomalies, however, show the familial patterns expected of multi-factorial inheritance. The aims and objective of this study were to study the incidence of visible congenital malformations at birth, to study risk factors, to find associated internal malformations.

**Methods:** It is a retrospective cross-sectional study carried out in a tertiary care hospital affiliated to a medical college. The Inclusion criteria include all new-borns delivered in the hospital with visible congenital malformations examined within 48 hours of birth. Extramural babies were included if they had presented within 48 hours after birth. The Exclusion criteria include still births were excluded from the study.

**Results:** Percentage of congenital malformation was 1.32%. Most common systems involved were musculoskeletal system (46.34%) followed by genitourinary system (21.34%) and gastrointestinal system (14.02%).

**Conclusions:** All Babies with gross congenital malformation should be screened for internal malformation. The incidence of CNS malformation has reduced than observed in previous studies which suggest awareness about antenatal folic acid supplementation. Other than CNS anomalies, other system anomalies were not diagnosed antenatally despite antenatal ultrasound being done in maximum number of mothers, which suggest use of 3D or 4D scan antenatally.

**Keywords:** Antenatal scan, Gross congenital malformation, New born

### INTRODUCTION

A congenital anomaly is a structural anomaly of any type that is present at birth. Congenital anomalies may be induced by genetic or environmental factors. Most congenital anomalies, however, show the familial patterns expected of multi-factorial inheritance. Indian people are living in the midst of risk factors for birth defects, e.g. universality of marriage, high fertility rate, large number of unplanned pregnancies, and poor coverage of antenatal care, poor maternal nutritional status and high rate of consanguineous marriages. Many

population-based strategies such as iodization, double fortification of salt, flour fortification with multivitamins, folic acid supplementation, periconceptional care, carrier screening and prenatal screening are some of proven strategies for control of birth defect.

According to March of Dimes (MOD) global report on birth defects, worldwide 7.9 million births occur annually with serious birth defects.<sup>1</sup> According to joint WHO and MOD meeting report, birth defects account for 7% of all neonatal mortality. In India, birth defect prevalence varies from 61 to 69.9/1000 live births.<sup>2</sup>

Congenital anomalies may be single or multiple and of minor or major clinical significance. About 3% of all live born infants have an obvious major anomaly.<sup>3</sup> Minor anomaly occur in higher percentage in newborn (10%).

The aetiology of approximately 50% of congenital anomalies is unknown of the remainder, etiology is attributed to 6-10% chromosomal, 3-7.5% single gene mendelian disorders, 20-30% multi-factorial and 6-7% environmental exposures. The development of more sensitive molecular technology has established etiology in many cases.<sup>4</sup>

In genetic causes, mendelian gene disorder and chromosomal abnormalities are included.<sup>5</sup> Environmental factors play an important role in causation of various congenital malformations. Some of them are drugs (thalidomide, alcohol, lithium etc.), maternal infections (rubella, toxoplasmosis, varicella, cytomegalovirus etc.), Hormones (maternal diabetes etc), Physical agents (X-ray, hyperthermia), regional factors (ectopic pregnancy, twins, amniotic band etc).<sup>3</sup> For preventive measure certain steps can be taken like, Ensuring adolescent girls and mothers have a healthy diet including a wide variety of vegetables and fruit, and maintain a healthy weight, ensuring mothers avoid harmful substances, particularly alcohol and tobacco, screening for infections, especially rubella, varicella, and syphilis, and consideration of treatment.<sup>6</sup>

**METHODS**

It was a cross-sectional observational study conducted at NICU of Shardaben Municipal General Hospital, affiliated to N.H.L. Medical College, Ahmedabad. The study period was from June 2017 to December 2018.

**Inclusion criteria**

- All newborns delivered in the hospital with visible congenital malformations examined within 48 hours of birth.
- Extramural babies were included if they presented within 48 hours after birth.

**Exclusion criteria**

- Still births were excluded from the study.

All the new-borns fitting the inclusion criteria were examined within 48 hours after birth. Relevant information regarding maternal age, antenatal status of mother, previous obstetric history, gestational age, sex, ethnicity, birth weight, birth order and consanguinity were documented.

Significant antenatal history like maternal illness, exposure to radiation, ingestion of drugs and complications of labour were noted. Antenatal ultrasonography findings were noted.

Ultrasound was carried in all new-borns except those with poor general condition. Where applicable, 2-D echocardiography, karyotyping and X-ray were done to detect internal anomalies associated with the visible defects. Review of antenatal USG was done.

Diagnosis of congenital anomalies was based on clinical evaluation of a new-born by a paediatrician soon after birth. Each patient received protocol-based management and advice.

**RESULTS**

A total 12,385 babies were delivered during the study period. Among them, 164 babies were born with congenital malformation. The overall percentage of babies with visible congenital malformation in the present study is 1.32% (Table 1).

**Table 1: Incidence of gross congenital malformations.**

	N
Total number of babies delivered	12385
Total number of malformed babies	164
Percentage	1.32

The relationship between maternal age and babies born with GCMF, in present study, revealed that a majority of such babies were born to mothers aged 20-29 years, which is not significant. This is comparable to study by Sarkar S et al.<sup>7</sup> Consanguineous marriages are reported to play a major role in the occurrence of GCA. It was observed in 10 parents. History of infections were found in 6 mothers (Table 2).

**Table 2: Maternal risk factors and malformations.**

	N
Age>35	4
Consanguinity	10
Infections	6
No antenatal care	7

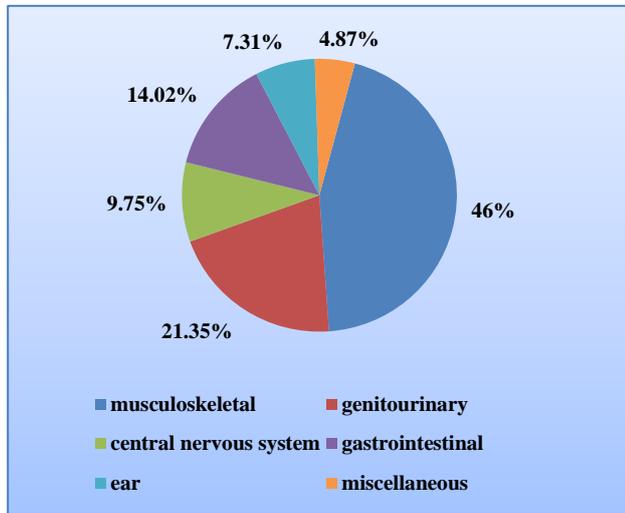
Associated internal anomalies were detected in 16 patients (9.75%). These were CHD, renal anomalies and CNS anomalies (Table 3).

**Table 3: Internal malformation associated with GCA.**

	N
Total babies with GCA	164
Associated internal anomalies	16
Not associated with internal anomalies	148

In the present study, musculoskeletal system was involved in 46%. Other systems which were involved are as following, genitourinary system in 21.35%, gastrointestinal system in 14.02%, central nervous system in 9.75%, ear in 7.31%, eye in 1.2% and miscellaneous in

4.87% (Figure 1). In present study, malformations were observed more amongst good weight babies. In >2.5 kg babies, percentage of malformation was 51.28%. In low birth weight babies, it was 48.17%.



**Figure 1: System wise distribution of malformation.**

The incidence of congenital malformation in a preterm baby was 2.15% where as in term babies was 1.2%.

In musculoskeletal system, CTEV was found in 31.09% of patients. Other anomalies which were found in musculoskeletal system are as following, polydactyly (8.53%), hypoplasia of digit (2.43%), Syndactyly (1.21%), Limb deformity (1.82%), Muscle defect (0.60%) and knee dislocation (0.60%).

In central nervous system, hydrocephalus and sacral dimple were equally found in 2.43% of patients. Meningomyelocele was found in 1.82%, Spina bifida in 1.21%, meningocele in 0.60%, microcephaly in 0.60% and encephalocele in 0.60% of patients.

In genitourinary system, hypospadias was found in 13.41% of patients. It was followed by cryptorchidism (4.87%), micropenis (1.82%), hydrocele (0.60%) and Chordee (0.60%). In gastrointestinal system, tracheoesophageal fistula was found in 6.70% of patients. Other anomalies found were cleft lip and palate (4.26%), gastroschis (1.21%), Inforated anus (1.21%), Oral cyst (0.60%).

## DISCUSSION

Total percentage of Congenital Malformation was 1.32%. It is comparable to other studies done in India. The incidence varied from 1.2%-2.5% in these studies.<sup>7-9</sup> Birth defects occur in approximately 3% of all live births and are the leading cause of infant mortality in U.S.<sup>10</sup>

Most common systems involved were musculoskeletal system (46.34%) followed by genitourinary system

(21.34%) and gastrointestinal system (14.02%). Musculoskeletal system was the most common system in various other studies conducted, which was followed by CNS anomalies.<sup>7-9</sup> In some other studies, CNS was the most common system involved which was followed by, musculoskeletal system.<sup>11,12</sup>

In musculoskeletal system maximum patients of congenital talipes equinovarus were found in the study (31.09%). This is comparable to other studies done by Vinodh SL et al, and study done by Sarkar S et al.<sup>7,8</sup> In it the percentage of CTEV was 15.3% and 17.1% respectively. Another study by Sandeep Sachdeva shows 17.02% CTEV with commonest anomaly being CNS.

Maximum patients of hypospadias were found in genitourinary system (13.4%). In study done by Vinodh SL et al, the incidence was 1.1% and in study done by Sarkar S et al, it was 1%.<sup>7,8</sup> All the babies in the present study were screened for electrolyte abnormalities and sugar. In one baby, hypoglycemia was detected, screening for congenital adrenal hyperplasia was done by dry blood spot. The first result showed positive for congenital adrenal hyperplasia, subsequently confirmatory screening was done which was found negative for CAH. Internal anomaly-cyanotic CHD (TOF) was detected in the same baby. Internal anomalies were also detected in another 2 babies, one had b/l inguinoscrotal hernia, and another had gross hydronephrosis with possibility of PUJ obstruction.

In GIT, maximum babies were of tracheoesophageal fistula (6.7%). The percentage of tracheoesophageal fistula in other studies were between 0.5-1.<sup>8,9</sup> In the present study, the percentage of gastroschis and imperforate anus is 1.21%. In study done by Sarkar S et al, the percentage of both anomalies was 0.7%. 4.26% of babies had cleft lip and palate in the study as compared to the study done by Vinodh SL et al, in which it was 2.4%.<sup>7,8</sup> A 4 had complete bilateral cleft lip palate and three had unilateral cleft lip palate.

In central nervous system, the most common anomaly found was sacral dimple and hydrocephalus (4 cases of each), followed by meningomyelocele (3 cases), spina bifida (2 cases) and meningocele, microcephaly, encephalocele (1 case of each). Hydrocephalus was also found to be maximum in other study (7.9%).<sup>8</sup> Hydrocephalus accounted to 1.7% in study done by Sarkar S et al.<sup>7</sup> Anencephaly was not found because stillbirths and abortions were not included and owing to high fatality rate of this condition, not a single live birth was found.

## CONCLUSION

All babies with gross congenital malformation should be screened for internal malformation. The incidence of CNS malformation has reduced than observed in previous studies which suggest awareness about antenatal folic acid supplementation. Other than CNS anomalies, other

system anomalies were not diagnosed antenatally despite antenatal ultrasound being done in maximum number of mothers, which suggest use of 3D or 4D scan antenatally.

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