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# Study of congenital malformations in fetus and its risk factors

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## ABSTRACT

**Aim:** Study of congenital malformations in fetus and its risk factors

Material and methods: Relevant information regarding maternal age, parity, gestational age, birth weight, sex, and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Whether the patient had an antenatal ultrasonography (USG) scan and findings were noted. All the aborted fetus and newborns were examined for congenital malformations soon after delivery.

**Results:** During the study period 40 congenital anomalies were seen in delivered babies and aborted foetuses, 28 (70%) patients were in the age group of 20-29 years, >30 years were 12 (30%). 31 (77.5%) patients had anomalies diagnosed on antenatal ultrasound. The number of congenital anomalies is more in low birth weight babies. The occurrence was more in female than male. Central nervous system was the most common system involved followed by musculoskeletal system. Anencephaly was the common malformation seen in 12 (30%) patients.

**Conclusion:** The most prevalent forms of CAs were abnormalities of the central nervous system and the musculoskeletal system at the time of birth. It is necessary to place restrictions on the prescription of drugs that have the potential to have a teratogenic impact in order to reduce the number of CAs.

**Keywords:** Congenital anomalies, Risk factors, Prevalence, Children/pediatric

## Introduction

Congenital anomalies are a major health problem and are responsible for a remarkable proportion of mortality and morbidity in newborns. In India the reported incidence is 2.5%. Congenital anomalies account for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India. [1] According to WHO Congenital anomalies are defined as structural or

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functional anomalies, including metabolic disorders which are present at the time of birth.[2] Around 40%- 60% of congenital anomalies are of unknown etiology.[3,4] 20-25% of anomalies the cause is multifactorial. 10-13% are because of environment and 12-25 % are attributed to genetic causes. Among the risk factors are advanced maternal and paternal age, consanguinity, teratogenic agents and nutritional deficiencies. Low socioeconomic status and poor antenatal care prevents early diagnosis of the malformations.

The World Health Organization (WHO) estimates that approximately three million babies are born each year, with major CAs accounting for approximately 3% of all newborns. [5] The global report of birth defects from 2006 showed that the prevalence of CAs varied between high-income countries, middle-income countries, and low-income countries, with middle-income and low-income countries accounting for 94% of all CAs. In Sudan, the CA rate was as high as 82 per 1,000 live births, while in France the rate was only 39.7 per 1,000 live births. [6] In 2006, the prevalence of CAs was between 45 and 50 per 1000 live births in the United States of America, the United Kingdom of Great Britain and Northern Ireland, and Germany. [7] The prevalence that was reported was significantly lower in both Africa and the Middle East. It ranged between 20 and 30/1000 live births in Kenya, Uganda, Nigeria, Saudi Arabia, and Pakistan. [8] A significant contributor to the morbidity and mortality of infants is the presence of congenital abnormalities. The World Health Organization (WHO) estimated that CAs were the cause of death for 11.3% of the world's 2.68 million neonatal deaths that occurred in 2006. Nearly all of the children who passed away as a result of CAs were residents of middle-income and low-income countries. [6] If not managed properly, congenital abnormalities can also result in long-term disabilities that affect a person's physical, mental, visual, and auditory capabilities. These disabilities can have significant adverse effects on individuals, families, the health care system, and societies. [5]

Our hospital is a tertiary care medical college hospital receiving patients from nearby districts. It is important to know the frequency, pattern of congenital anomalies and various presentations. This in turn will help to develop strategies for patient counselling and management. Patient should be seen early in pregnancy and second trimester ultrasound scan should be performed. Neonatal management along with medical and surgical intervention counselling needs to a part of the strategy. Obstetric management planning will prevent complications as these patients have associated risk factors like anaemia, gestational diabetes, polyhydramnios, and malpresentations.

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This study was aimed at presenting the spectrum of various congenital anomalies, epidemiological features of pregnant women with anomalous fetus. Fetal and neonatal details. Other associated antenatal complications and mode of delivery.

**Material and methods**

This Cross sectional study was done on 40 antenatal patients who delivered or aborted congenital anomalous baby from a period of 1st JULY 2020 to 31st August 2022 admitted at OBGY department at Sri Aurobindo Medical College and Post Graduate Institute, Indore and who satisfy the inclusion criteria was included in this study. Informed written consent was taken. A pre-structured proforma was used to collect the baseline data. Detailed clinical examination and biochemical tests will be done on all patients as per the protocol. Relevant information regarding maternal age, parity, gestational age, birth weight, sex, and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Whether the patient had an antenatal ultrasonography (USG) scan and findings were noted. All the aborted fetus and newborns were examined for congenital malformations soon after delivery.

**Inclusion Criteria**

- Pregnant women with Target scan and NT scan reports with radiological findings suggestive of congenital malformations

**Exclusion Criteria**

- Women who do not give consent to participate in this study

**Statistical Analysis**

Sample size of the study was 40. Descriptive statistics will be used to show the features and characteristics of the collected data. Chi square test will be applied on categorical data P value <0.05 will be taken as significant.

**Results**

During the study period 40 congenital anomalies were seen in delivered babies and aborted foetuses, 28 (70%) patients were in the age group of 20-29 years, >30 years were 12 (30%). 17 (42.5%)patients were primigravida. 35 (87.5%) patients were from rural area and 29 (72.5%) were unbooked patients. Majority of patients 32 (80%) had only middle school education and were from low socio economic status. The incidence of congenital malformations was higher among abortions and preterm deliveries with respect to full term

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deliveries (26). The number of still births was high in this group (85%). 31 (77.5%) patients had anomalies diagnosed on antenatal ultrasound. 14 patients who did not have antenatal scan 8(50%) underwent caesarean section. The indications were neonatal like IUGR, fetal, and distress.

Table 1. Demographic profile of the patients

| Age | number | % |
|---|---|---|
| 20-29 | 28 | 70 |
| >30 | 12 | 30 |
| Gravida | | |
| Primigravida | 17 | 42.5 |
| Multigravida | 23 | 57.5 |
| Area | | |
| Rural | 35 | 87.5 |
| Urban | 5 | 12.5 |
| Booking status | | |
| Unbooked | 29 | 72.5 |
| Booked | 11 | 27.5 |

Table 2. Anomalies diagnosed on antenatal ultrasound

| | | |
|---|---|---|
| anomalies diagnosed on antenatal ultrasound | 31 | 77.5 |

Table 3. Congenital anomalies

| Birth weight | Number | Percentage |
|---|---|---|
| low | 30 | 75 |
| Average | 10 | 25 |
| Sex | | |
| Male | 15 | 37.5 |
| Female | 25 | 62.5 |

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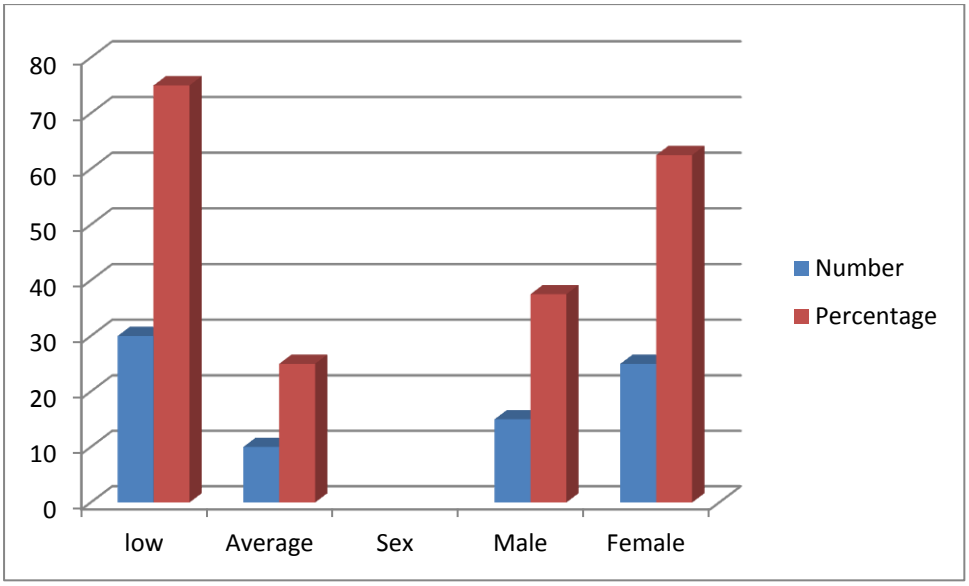


Fig 1. Congenital anomalies

The number of congenital anomalies is more in low birth weight babies. The occurrence was more in female than male. Central nervous system was the most common system involved followed by musculoskeletal system Table 4. Anencephaly was the common malformation seen in 12 (30%) patients. One patient with consanguineous marriage, had history of congenital malformation in previous pregnancy. Despite all antenatal investigations in current pregnancy had Epidermolysis Bullosa (undiagnosed) with intrauterine fetal death at 37 weeks of gestation. She had a caesarean section delivery for non-progress of labour.

Table 4. System involved in congenital anomaly

| System involved in congenital anomaly | Number | Percentage |
|---|---|---|
| Digestive system anomalies | 3 | 7.5 |
| Musculoskeletal anomalies | 12 | 30 |
| Circulatory system anomalies | 3 | 7.5 |
| Genetic disorders | 2 | 5 |
| Central nervous system | 20 | 50 |

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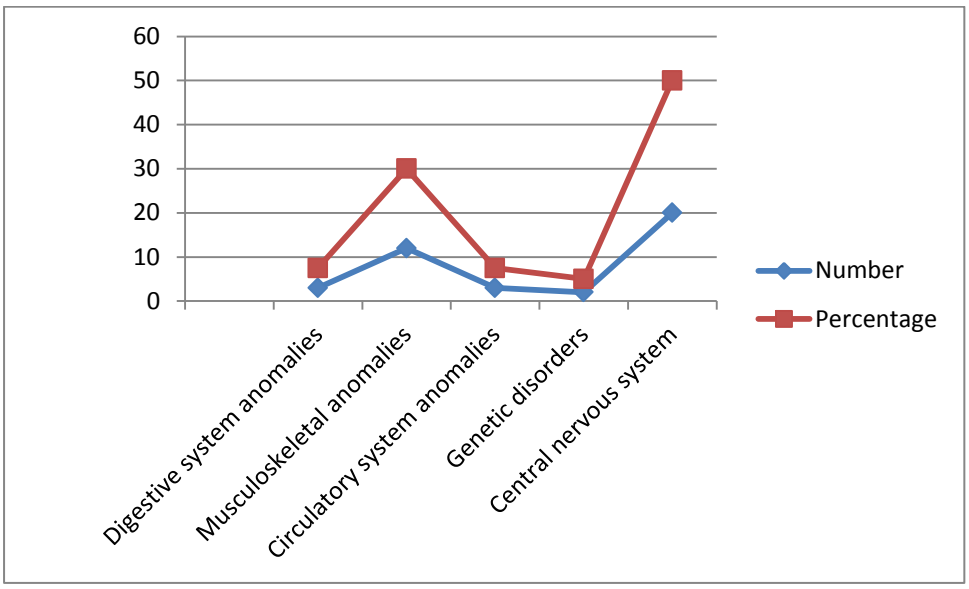


Fig 2. System involved in congenital anomaly

**Discussion**

This percentage doubles in the first year and reaches 10% by the age of 5 years, as more birth problems become apparent and can be detected as the child gets older. Two to three percent of all babies are discovered to have birth defects. Birth malformations are the underlying cause of over twenty percent of all infant fatalities.[9] The occurrence of congenital anomalies seen in delivered babies and aborted foetus was more prevalent in primigravida and in the age group of 20-29 years in 28 (70%) patients, which differs from the findings of other studies, which found that a higher incidence was observed in babies born to mothers with an age greater than 30 years. Singh A and others [10] Both of these findings might be explained by the fact that 77% of patients came from rural areas and 65% of patients were from poor socioeconomic backgrounds and did not get any prenatal care. [6]

In the present research, abnormalities of the digestive system made up 7.5% of the overall CAs, whereas abnormalities of the musculoskeletal system made up 30% of the total CAs. They were higher than those that were reported in a surveillance study that was carried out in Glasgow and Clyde in 2015–2016. In that research, GIT abnormalities only formed 9% of the total, whereas musculoskeletal anomalies constituted 25% of the total. [11] The current investigation was a hospital-based case series, while the other study used a population-based surveillance programme; this might explain why the two studies came to different conclusions about the incidence rates.

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In this particular research, abnormalities pertaining to the cardiovascular system made up 7.5% of the total CAs. According to the results of the surveillance research that was carried out in Glasgow and Clyde, the prevalence of circulatory system abnormalities was determined to be 13.9% [11]. The data from EUROCAT indicated even higher rates for circulatory system anomalies, which were 35%. [12] It's possible that the disparity in rates is attributable to different approaches of data collection as well as varying degrees of exposure to different kinds of hazards.

According to the results of the case-control research, the likelihood of females developing CAs was shown to be 1.67 times greater than that of men. This conclusion was consistent with the findings of Tennant et al., who discovered a 15% greater incidence of CAs in female babies in North England compared to male infants in the same region (1985–2003). [13]

According to the findings of the present research, children born to mothers older than 30 years had a higher prevalence of CA than children born to mothers younger than 30 years, however this difference did not reach statistical significance. Two separate studies, one conducted in Tanzania in 2013 [14] and the other at Ain Shams University in Cairo between 1995 and 2009 [15], found a statistically significant correlation between mothers older than 35 years old and CAs.

## Conclusion

The most prevalent forms of CAs were abnormalities of the central nervous system and the musculoskeletal system at the time of birth. It is necessary to place restrictions on the prescription of drugs that have the potential to have a teratogenic impact in order to reduce the number of CAs.

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