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A Retrospective Assessment of the Occurrence of Congenital Anomalies in Intrauterine Foetus of 20–24 Weeks of Pregnancy: A Hospital-Based Study

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ABSTRACT

Background: Congenital anomalies are an important cause of perinatal and neonatal mortality and long-term morbidity, particularly in developing countries. Early detection during the second trimester is crucial for counselling and management. The present study was undertaken to assess the occurrence of congenital anomalies in intrauterine foetuses of 20–24 weeks of gestation.

Aim: To assess the occurrence and pattern of congenital anomalies in intrauterine foetuses of 20–24 weeks of pregnancy and to evaluate the associated maternal risk factors.

Materials and Methods: This was a retrospective, analytical, hospital-based study of patients who delivered or aborted a congenitally anomalous baby. During the study period, 50 congenital anomalies were identified among delivered babies and aborted foetuses. Relevant demographic, obstetric and clinical information was documented and analysed.

Results: Of the 50 patients, 37 (74%) were aged 20–29 years and 12 (24%) were >30 years; 18 (36%) were primigravida. Thirty-nine (78%) patients belonged to a rural area and 32 (64%) were unbooked. Antenatal ultrasonography detected anomalies in 35 (70%) patients, while 15 (30%) had not undergone an antenatal scan. Sixteen (32%) patients underwent lower segment caesarean section (LSCS) and 18 (36%) were preterm. The principal associated maternal risk factors were anaemia (30%), polyhydramnios (20%), previous caesarean section (20%), oligohydramnios (16%), pregnancy-induced hypertension (16%), twins (10%), breech presentation (10%), intrauterine growth restriction (10%) and Rh-negative status (10%).

Conclusion: Congenital anomalies were more common among rural, unbooked patients and were associated with a higher rate of stillbirth and preterm delivery. Early diagnosis through routine antenatal ultrasonography and appropriate genetic counselling for the index and subsequent pregnancies are essential for the proper management of this important public health problem.

Keywords*: Congenital anomalies; intrauterine foetus; second trimester; antenatal ultrasonography; perinatal mortality.*

INTRODUCTION

The term "congenital" denotes a condition that is *present at birth*. Congenital malformation refers to errors in morphogenesis occurring during early foetal development[1] and is characterised as physical, metabolic or anatomical abnormalities that may be detected before birth, at birth or during the first year of life.[2] Congenital abnormalities make a significant contribution to foetal and neonatal mortality and impairment globally[3], accounting for 8–15% of perinatal deaths and 13–16% of newborn deaths in India.[4,5] Perinatal mortality is one of the most sensitive indicators of the quality of maternal and neonatal care.[6] In recent years, congenital diseases have emerged as a major public health problem in

developing nations, attributable to a relative decline in deaths from infections and malnutrition and a corresponding rise in morbidity and mortality from congenital anomalies.[7]

Ultrasound is a crucial diagnostic tool for the identification of numerous developmental abnormalities. The initial diagnostic ultrasound screening is generally conducted between 11 and 13 weeks of gestation, during which nuchal translucency thickness is assessed and the presence of the nasal bone is verified.[8,9] Birth defects (BDs) or congenital anomalies are defined as anatomical or functional abnormalities, including metabolic disorders, that arise during intrauterine development—predominantly within the first trimester of gestation.[10–12] The World Health Organization (WHO) reports that BDs are among the leading contributors to spontaneous abortion, stillbirth and mortality and impairment in children under five years of age. Approximately 3–6% of newborns are born with severe birth defects, and over 3.3 million children succumb to birth defects globally each year.[13,14]

Anomaly scans have become standard practice during the prenatal period, with the optimal time for detection being approximately 18 weeks of gestation. Some defects may, however, evade detection by conventional ultrasonography and necessitate a foetal autopsy to identify accompanying anomalies and confirm the diagnosis. This aids in identifying the cause of foetal loss and is therefore an important component of subsequent genetic counselling.[15] Central nervous system (CNS) abnormalities are among the most prevalent categories of congenital malformations.[16,17] A considerable proportion of congenital defects cannot be identified on prenatal assessments such as ultrasonography alone, necessitating autopsy.[18] Foetal autopsy is commonly indicated in cases of congenital abnormality, intrauterine foetal demise and inevitable abortion.[19]

The objective of the present study was to evaluate the prevalence and pattern of congenital abnormalities in intrauterine foetuses at 20–24 weeks of gestation in a tertiary care setting.

MATERIALS AND METHODS

Study design and setting: This was a retrospective, analytical, hospital-based study conducted in the Department of Anatomy in collaboration with the Department of Obstetrics and Gynaecology, MGM Medical College & LSK Hospital, Kishanganj, Bihar.

Study population: Patients who delivered or aborted a baby with a congenital anomaly during the study period were included. A total of 50 congenital abnormalities were identified among delivered babies and terminated pregnancies.

Data collection: Pertinent information was gathered for each case, including the mother's age, parity, gestational age, birth weight, sex of the baby and history of consanguinity. A detailed antenatal history was recorded, including the presence of maternal disease, intake of medications, exposure to radiation and complications related to delivery. Where the patient had undergone antenatal ultrasonography (USG), the findings of the scan were documented. Each newborn and terminated foetus was examined shortly after delivery for the presence of congenital abnormalities, and the findings were correlated with the antenatal scan reports where available.

Statistical analysis: Data were tabulated and expressed in terms of frequency and percentage. Categorical variables were summarised in tabular form to identify the demographic pattern, mode of delivery, antenatal detection rate and associated maternal risk factors.

RESULTS

A total of 50 cases of congenital anomalies were analysed. The demographic distribution of the patients is presented in Table 1. The majority of mothers (74%) belonged to the 20–29-year age group, were multiparous (64%) and resided in rural areas (78%); nearly two-thirds (64%) were unbooked at the time of presentation.

Table 1. Demographic pattern of patients (n = 50)

Variable	N	%	Variable	N	%
Age (years) — <19	1	2	Residence — Rural	39	78
20–29	37	74	Urban	11	22
>30	12	24	Booking — Booked	18	36
Parity — Primi	18	36	Unbooked	32	64
Multi	32	64	—	—	—

Antenatal ultrasonography detected congenital anomalies in 35 (70%) of the 50 patients, while 15 (30%) had not undergone an antenatal scan during pregnancy (Table 2).

Table 2. Antenatal ultrasonographic detection of congenital anomalies

Antenatal scan finding	N	%
Patients with anomalies diagnosed on USG	35	70
Patients with anomalies not diagnosed on USG	15	30

The mode of delivery is summarised in Table 3. Sixteen (32%) patients underwent lower segment caesarean section (LSCS) and 18 (36%) delivered preterm; 11 (22%) cases ended in abortion.

Table 3. Mode of delivery

Mode of delivery	N	%
Abortion	11	22
Preterm delivery	18	36
Vaginal delivery	5	10
Lower segment caesarean section (LSCS)	16	32

The various maternal risk factors associated with congenital anomalies in the present study are detailed in Table 4. Anaemia was the most frequent risk factor (30%), followed by polyhydramnios (20%) and a history of previous caesarean section (20%).

Table 4. Associated maternal risk factors

Risk factor	N	%
Anaemia	15	30
Oligohydramnios	8	16
Polyhydramnios	10	20
Breech presentation	5	10
Twin pregnancy	5	10
Preterm delivery	28	56
Pregnancy-induced hypertension (PIH)	8	16
Rh-negative status	5	10
Previous caesarean section	10	20
Intrauterine foetal death (IUFD)	6	12

DISCUSSION

Congenital abnormalities are a significant health issue and account for a substantial proportion of mortality and morbidity in neonates. They affect 3–5% of live births in the United States, while the reported frequency in India is approximately 2.5%. Congenital abnormalities constitute 8–15% of perinatal deaths and 13–16% of newborn deaths in India.[20] The WHO defines congenital anomalies as structural or functional abnormalities, including metabolic disorders, that are evident at birth.[21] Approximately 40–60% of congenital abnormalities have an unidentified aetiology.[22,23]

In the present study, 37 (74%) patients were aged 20–29 years, while 12 (24%) were over 30 years of age, and 18 (36%) were primigravida. Thirty-nine (78%) patients resided in rural areas and 32 (64%) were unbooked. Previous studies have similarly demonstrated that the incidence of congenital abnormalities is elevated among abortions and preterm deliveries, and the findings of Bhat et al. and Sachdeva et al. are consistent with those of our investigation.[24–26] In our cohort, 70% of patients had defects identified on prenatal ultrasonography, whereas 15 patients had not undergone an antenatal scan. Thirty-two per cent underwent LSCS, and 36% delivered preterm. The study identified several associated risk factors—anaemia (30%), oligohydramnios (16%), polyhydramnios (20%), twins (10%), breech presentation (10%), intrauterine growth restriction (10%), previous caesarean section (20%), Rh-negative mother (10%) and pregnancy-induced hypertension (16%). A similar pattern of risk factors was reported by Gupta et al.[27]

Congenital abnormalities are a primary cause of foetal demise, and autopsy considerably aids in the detection of intrauterine foetal death.[28] The family requires accurate information regarding the aetiology of foetal demise, as the couple's future reproductive choices are contingent upon the underlying cause; this also allows prediction of recurrence risk and may help avert analogous losses in subsequent pregnancies.[29] Identifying defects and deformities prenatally through different investigations such as ultrasonography, chromosomal analysis, foetal autopsy and placental evaluation mitigates the chance of recurrence, which may otherwise reach 25%.[30]

CONCLUSION

The findings of the present study indicate that congenital abnormalities were more frequent among patients from rural areas and were associated with a higher incidence of stillbirth and preterm delivery. To effectively manage this important public health problem, early diagnosis through routine prenatal ultrasonography, comprehensive antenatal care for all pregnant women—including those from rural and unbooked populations—and appropriate counselling for both the index pregnancy and any subsequent pregnancies are essential.

LIMITATIONS OF THE STUDY

The principal limitations of this study are its retrospective design, the relatively small sample size of 50 cases and the fact that it was conducted at a single tertiary care centre. Karyotyping and detailed genetic analysis could not be performed in all cases. Larger, prospective, multicentre studies incorporating cytogenetic evaluation are recommended to confirm and extend the present findings.

CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

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None.

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