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Autopsy-Based Assessment of Congenital Malformations in Fetal and Perinatal Deaths

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ABSTRACT

Background: Congenital malformations are a major cause of fetal and perinatal mortality in developing countries. Despite advances in prenatal diagnosis, many anomalies remain undetected, making fetal autopsy essential for accurate evaluation.
Methods: This combined retrospective and prospective autopsy-based study was conducted in the Department of Pathology, KMCRI, Hubballi, from July 2017 to June 2020. Among 400 fetal autopsies, 120 cases with congenital anomalies (20 weeks of gestation to 7 days postnatal life) were analysed. Detailed gross and histopathological examinations were performed, and maternal and fetal variables were assessed descriptively.
Results: Congenital malformations were identified in 34% of cases. Most fetuses were 20–24 weeks of gestation (55.8%) and weighed 350–1000 g (79.2%). Male predominance was noted (56.7%). Central nervous system anomalies were most frequent (47.5%), followed by musculoskeletal (28.3%) and genitourinary (23.3%) defects. Anencephaly and meningomyelocele were the commonest lesions. Multiple system involvement occurred in 27% of cases.
Conclusion: Fetal autopsy remains the gold standard for detecting and characterising congenital anomalies and is indispensable for determining the cause of death and guiding future genetic counselling.

Keywords: Congenital anomalies, Fetal autopsy, Perinatal mortality.

INTRODUCTION

Inherent irregularities, or congenital anomalies, represent structural or functional deviations from normal development, including metabolic disorders, that arise during intrauterine life and may be detected prenatally, at birth, or later in life[1]. These abnormalities may result from single-gene mutations, chromosomal defects, multifactorial inheritance, exposure to teratogens, or micronutrient deficiencies[1]. They constitute a significant cause of perinatal morbidity and mortality worldwide, accounting for approximately 8–15% of perinatal deaths and 13–16% of neonatal deaths in India[2]. In recent years, congenital anomalies have emerged as a major public health concern in many developing countries. This shift is largely attributable to improved control of infectious diseases and nutritional deficiencies, leading to a relative increase in

deaths due to congenital malformations[3]. Although prenatal screening methods such as ultrasonography and maternal serum testing are valuable, a substantial proportion of anomalies remain undetected, underscoring the continued importance of fetal autopsy[4]. Autopsy not only confirms antenatal diagnoses but also frequently identifies additional or unsuspected abnormalities[3]. Despite advances in medical care, fetal and perinatal autopsy remains the most reliable method for determining the cause of death and plays a crucial role in prenatal genetic counseling[5]. It is estimated that 2–3% of live-born infants have a major congenital anomaly, and approximately 0.7% have multiple serious malformations[6]; however, the true prevalence is likely higher, as many severely affected fetuses result in spontaneous pregnancy loss[6]. The present study aims to evaluate the frequency and spectrum of congenital anomalies in perinatal deaths and to analyse their clinicopathological features, to improve early diagnosis, management, and preventive strategies in future pregnancies.

MATERIALS & METHODS

Study Design and Data Source:

This autopsy-based, combined retrospective and prospective study was conducted in the Department of Pathology, KMCRI, Hubballi, over a period of three years (July 2017–June 2020).

Method of Data Collection:

All autopsy examinations were conducted after obtaining written informed consent from the parents or legal guardians. In cases where congenital anomalies were suspected clinically or on radiological evaluation, informed consent was obtained, and the fetus or relevant samples were referred through the Department of Obstetrics and Gynaecology for further pathological examination. A single pathologist conducted all examinations using a uniform and systematic protocol, with careful review of clinical records and imaging findings, followed by detailed external and internal examination of the fetus. The study was approved by the Institutional Ethics Committee and was conducted in accordance with the Declaration of Helsinki and its subsequent revisions (IEC No. 150/2018; Date: 15/11/2018).

Sample Size:

A total of 120 fetuses with congenital anomalies were included, comprising one year of retrospective data and two years of prospective observations.

Inclusion and Exclusion Criteria:

All fetuses from 20 weeks of gestation up to 7 days of postnatal life were included. Severely autolysed specimens were excluded.

Histopathological Examination:

After evisceration, individual organs were weighed, and representative tissue samples were taken from the brain, heart, lungs, liver, kidneys, spleen, adrenal glands, colon, placenta, and umbilical cord. The specimens were processed and stained using standard histopathological techniques for microscopic evaluation.

Statistical Analysis

Data were tabulated and analysed to assess associations between congenital anomalies and maternal as well as fetal variables, including maternal age, parity, previous obstetric history, fetal birth weight, gestational age, and sex. Results were expressed as frequencies and percentages using Microsoft Excel 2021.

RESULT

The present study was a one-year retrospective review and a two-year prospective study conducted over three years in the Department of Pathology, KMCRI, Hubballi. A total of 120 cases of congenital anomalies were identified through fetal post-mortem examination within the study, meeting the inclusion criteria.

A total of 120 fetuses with congenital anomalies were included in the study. The majority of cases were observed between 20–24 weeks of gestation (55.8%), followed by 25–29 weeks (25.8%). Most fetuses had a birth weight of 350–1000 g (79.2%). Male fetuses predominated (56.7%) as shown in Table 1 below.

Table 1: Fetal demographic factor

Fetal Demographic Factors		No. of cases	Percentage (%)
Gestational age (in weeks)	20-24	67	55.8
	25-29	31	25.8
	30-34	13	10.9
	35-39	9	7.5
Birthweight (grams)	350- 1000	95	79.2
	1001-2000	23	19.2
	2001-3000	2	1.6
Gender	Male	68	56.7
	Female	52	43.3

Table 2: Maternal Demographic Factors

Maternal Demographic Factors		No. of cases	Percentage (%)
Maternal age (years)	19	6	5.0
	20-24	78	65.0
	25-29	30	25.0
	30-34	6	5.0
Parity	Primigravida	58	48.3
	Multigravida	62	51.7
History of previous abortion	Present	20	16.7
	Absent	100	83.3

Among the mothers, the most common age group was 20–24 years (65.0%), followed by 25–29 years (25.0%). Mothers aged 19 years and those between 30–34 years each accounted for 5.0%. Parity was nearly evenly split, with multigravida women making up 51.7% and primigravida 48.3%. A history of previous abortion was reported in 16.7% of mothers, while the majority (83.3%) had no such history.

Table 3: System-wise Distribution of Congenital Anomalies

System	Major Anomalies	Number	Percentage (%)
Central Nervous System (n = 57)	Anencephaly	18	31.5
	Meningomyelocele	17	29.8
	Arnold–Chiari malformation type II	6	10.5
	Congenital hydrocephalus	5	9.4
	Others*	11	19.3
Musculoskeletal System (n = 34)	Diaphragmatic hernia	7	20.5
	CTEV	7	20.5
	Omphalocele	5	14.7
	Skeletal dysplasia	5	14.7
	Others*	10	29.6
Genitourinary System (n = 28)	Multicystic renal dysplasia	15	53.6
	Renal agenesis	9	32.1
	Polycystic kidney disease	4	14.3
Cardiovascular System (n = 10)	Hypoplastic left heart syndrome	5	50.0
	Others*	5	50.0
Gastrointestinal System (n = 4)	Imperforate anus	2	50.0
	Others*	2	50.0
Respiratory System (n = 3)	Congenital Adenomatoid Malformation	1	33.33
	Absent Lung	1	33.33
	Hypolobation	1	33.33

Central nervous system anomalies were the most frequent (47.5%), followed by musculoskeletal (28.3%) and genitourinary (23.3%) systems. Multiple system involvement was noted in 27% of fetuses.

FIGURE 1: Anencephaly

FIGURE 2: Encephalocele

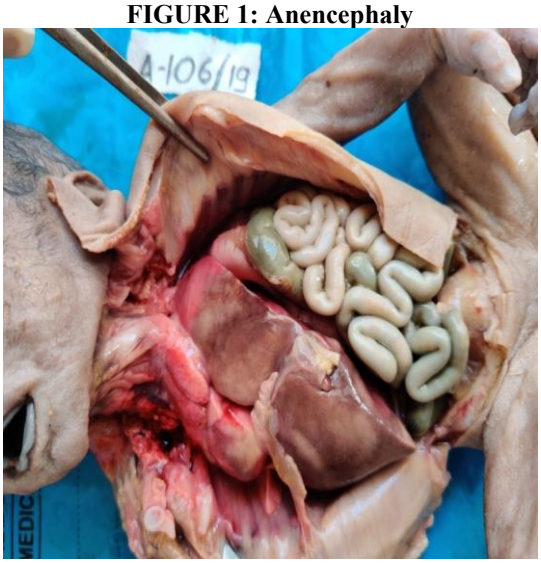

FIGURE 3: Congenital diaphragmatic hernia: left diaphragmatic defect causing herniation of liver, spleen and bowel loops.

FIGURE 4: Sirenomelia, showing fusion of both the lower limbs.

DISCUSSION

Congenital malformations remain a major cause of perinatal mortality worldwide. In developed countries, they account for approximately 25–30% of perinatal deaths, whereas in developing nations such as India, they contribute to about 10–15%[7,8]. With improvements in maternal health services, control of infectious diseases, and better nutritional status, the relative contribution of congenital anomalies to fetal and neonatal loss has become increasingly evident. Consequently, attention has shifted toward early detection and prevention of these conditions. The embryonic period, particularly between the 3rd and 8th weeks of gestation, represents the most critical phase, during which both genetic and environmental factors, including infections and teratogenic exposures, can adversely affect normal development. In this context, fetal autopsy plays a pivotal role in confirming antenatal diagnoses, elucidating the exact nature of anomalies, and guiding recurrence risk assessment and reproductive counseling[1].

In the present study, 400 fetal autopsies were performed over three years, of which 120 (34%) revealed congenital malformations. This prevalence is comparable to that reported by Faye-Petersen et al[9] (34.5%) and Andola et al[10] (44%), and exceeds the 25.5% observed by Kale et al[11]. The majority of affected mothers were in the 20–24-year age group, in agreement with Kale's observations, but differing from studies by Bhatia et al[12] and Siva Sankara Naik et al[13], who reported higher maternal age predominance.

A slightly higher incidence of anomalies was observed among multigravida women (51%), similar to the findings of Bhatia et al[12], whereas Naik et al[13] documented a predominance among primigravidas. Most mothers (80%) had no history of previous abortions, consistent with Kale et al[11]. The majority of affected fetuses weighed between 350–1000 g (79.2%) and were delivered between 20–24 weeks of gestation (55%), a pattern differing from studies reporting later gestational presentation. A male predominance was noted in our series, in contrast to some previous reports showing female preponderance.

System-wise analysis demonstrated a predominance of central nervous system anomalies (47.5%), followed by musculoskeletal (28.3%) and genitourinary (23.3%) defects. This distribution parallels observations by Andola[10], Kapoor[14], and Tomatir[15], although Kale[11] and Prabhala[16] reported higher musculoskeletal involvement. The higher prevalence of central nervous system anomalies may be attributed to maternal drug intake during early pregnancy, leading to defective neural tube formation. Anencephaly and meningomyelocele were the most frequent CNS malformations, similar to other Indian studies.

Cardiovascular anomalies exhibited varied morphology, with hypoplastic left heart syndrome being the most common. Respiratory anomalies were infrequent and included rare entities such as pulmonary agenesis and hypolobation, while gastrointestinal defects primarily comprised imperforate anus, jejunal atresia, and polysplenia. Omphalocele was classified under musculoskeletal anomalies in accordance with updated WHO recommendations.

Genitourinary malformations were dominated by multicystic renal dysplasia, followed by polycystic kidney disease and renal agenesis. Musculoskeletal anomalies mainly included limb defects and diaphragmatic hernia. Syndromic associations, including Down syndrome, Patau syndrome, Pena–Shokeir syndrome, and VACTERL association, were identified, although cytogenetic confirmation was not available, representing a limitation.

Multiple system involvement was noted in 27% of cases, comparable to the findings of Kapoor and Kale, and higher than that reported by Andola et al[10]. This emphasizes the indispensable role of comprehensive fetal autopsy in accurately identifying complex malformation patterns and improving genetic counselling and preventive strategies.

Limitations

The primary limitations of this study is constraints in resources; advanced genetic and chromosomal analyses could not be performed. As a result, the precise etiological diagnosis in some cases could not be established.

CONCLUSION

Congenital malformations remain one of the leading causes of fetal and neonatal deaths, as well as long-term disabilities across the globe. In recent years, they have emerged as a significant public health concern in developing countries, where a substantial proportion of these anomalies are lethal. Perinatal autopsy plays a vital role in detecting and confirming these malformations. Beyond confirming clinical suspicions, it serves as a powerful tool for quality assurance in diagnosis, often uncovering findings that may have been missed during routine prenatal screening.

While prenatal ultrasonography has advanced considerably and is useful in identifying many fetal anomalies, autopsy remains the gold standard. It can reveal additional or unsuspected abnormalities, refine or change clinical diagnoses, and offer a more comprehensive understanding of the cause of death. A thorough autopsy allows accurate identification and classification of visceral malformations, providing critical insights not just for academic knowledge but for real-world clinical application. Importantly, the findings from fetal autopsy help clinicians counsel families regarding recurrence risks and guide future reproductive planning, making it an essential component of perinatal care.

Declaration by Authors
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