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A Retrospective Study of Fetal Congenital Anomalies In Tertiary Care Hospital

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Abstract

Background:
Congenital anomalies are a significant cause of fetal morbidity and mortality. Fetal autopsy remains the gold standard for confirming congenital malformations and assessing recurrence risk.

Methods:
A retrospective review was conducted on fetal autopsy at a tertiary care hospital over a defined period. Cases with confirmed congenital anomalies were analysed for prevalence, type of anomaly and associated clinical details. Anomalies were categorized system-wise to identify the most commonly affected systems.

Results:
Out of 264 fetal autopsies examined, congenital anomalies were observed in 100 cases (37%). Central nervous system malformations were the most prevalent, followed by musculoskeletal, cardiovascular, and renal anomalies. Multiple system involvement was also noted.

Conclusion:
Fetal autopsy pathology is crucial for accurate diagnosis, guiding genetic counseling and preventing recurrence through targeted interventions and improved antenatal care.

Keywords: *Congenital, Anomalies, Fetal, Autopsy, Anencephaly*.

1. INTRODUCTION

Congenital anomalies are structural or functional anomalies present at birth that can cause physical disability, intellectual and developmental disabilities (IDDs), and other health problems.1 Congenital anomalies may be hereditary or sporadic, isolated or multiple, gross or microscopic. These birth defects arise from multiple factors including genetic disorders, poor maternal nutrition, TORCH infections, alcohol use, environmental toxins (pesticides, tobacco, pollutants), maternal diseases, and advanced maternal age.2 Recent population-based study (1969–2024) estimated the global prevalence of major congenital anomalies at 0.86 to 3.11 cases per 10,000 births.3 A total of 42 cases of major congenital anomalies were recorded, with an overall prevalence of 230.51 per 10,000 births in first cohort study done in India.4 As per the

literature search in year 2015, meta-analysis estimated up to 472,177 congenital anomaly-affected births in India annually with anencephaly and talipes being the most common anomalies.[5]

The meticulous performance of fetal autopsies plays a critical role in the comprehensive evaluation of congenital anomalies. A thorough examination, including detailed gross and histopathological assessments, genetic analysis, and imaging techniques, helps identify structural abnormalities, metabolic disorders, and syndromic conditions that may not have been diagnosed prenatally.

There is limited data on the prevalence and morphological characters of congenital malformations in our region, highlighting the need for further research and surveillance.

Understanding the prevalence and classification of fetal anomalies is crucial for improving prenatal diagnosis, guiding clinical management, and developing targeted preventive strategies. Prevalence data helps to identify trends and assess risk factors. Classifying fetal anomalies by organ system improves diagnosis, guides treatment, aids parental counselling and supports research. This study aims to estimate prevalence of congenital anomalies.

2. MATERIALS AND METHODS:

This retrospective study was carried out in tertiary care hospital in South India for a period of 2 years from 1st June 2021 to 31st May 2023 after obtaining institutional ethical clearance (Reference No. MDC/JNMCIEC/257). Our study included all fetuses received for autopsy after obtaining parental consent. Maternal clinical history, demographic data and available radiological findings were collected from Medical Record Department and the autopsy was carried out following standard procedure

For optimal fixation 10% formalin was injected into the abdominal cavity, thorax (right and left side) and brain. After injecting, the fetus was stored in 10% formalin for 8-10 hours. If skeletal defects were suspected, an infantogram was done prior to fixation.

Detailed external and internal examination was carried out assessing different parameters. (Table 1)-

External examination	**Internal Examination**
Body weight	Cranial cavity
Circumferences- Head/Abdomen/Chest	Thoracic cavity
Crown rump length, Rump heel length, Crown heel length	Abdominal cavity
Hand and foot length	Systemic description- Major organs
Maceration	
Meconium staining	
Any congenital anomaly	

Table 1- Parameters for autopsy examination

Detailed histopathological examination was carried out for all the fetuses. At least 1 block of all major thoracic and abdominal organs was submitted. Photographs of gross examination was taken for documentation. The anomalies were further classified as per WHO international classification of diseases.[6]

3. RESULTS

Among the 264 fetuses examined in this study, congenital anomalies were present in 37% of cases, with central nervous system defects emerging as the most prevalent type of malformation.

Most anomalous fetuses in this study were male (55%). Gestational age ranged from 14 to 40 weeks. Majority were second trimester abortions [68%] with 21 % being first trimester and 11% being third trimester abortions. The majority of mothers (46%) belonged to age group 26–30 years, followed by 26% each in the <20 and 21–25 age groups and only 6% are over 30 years. Fourteen percent fetuses (n=100) had prenatal USG findings; out of which 12 % USG findings correlated with the autopsy examination findings. The infantogram was done in approximately 11% cases of suspected skeletal anomaly.

External examination parameters	Mean	Range
Weight of Fetus	367.31 grams	10-1300 grams
Crown Heel Length	29.15 cm	6-44 cm
Crown Rump Length	17.65 cm	4-28 cm
Head Circumference	17.53 cm	2-29 cm
Chest Circumference	15.47 cm	2-26 cm
Abdominal Circumference	15.77 cm	2-26 cm
Hand Length	2.61 cm	0.5-8 cm
Foot Length	3.35 cm	0.5-8.5 cm

Table 2: External examination parameters with mean value

The external examination of 100 fetuses revealed an average weight of 367.31 grams. The mean crown-heel and crown-rump lengths were 29.15 cm and 17.65 cm, respectively. Head, chest, and abdominal circumferences were fairly consistent, averaging 17.53 cm, 15.47 cm, and 15.77 cm. The mean hand and foot lengths were 2.61 cm and 3.35 cm, providing key reference points for fetal growth assessment.

In anomalous male fetuses, majority (43%) showed CNS anomaly and in female fetuses, circulatory system anomalies were predominantly observed (23%). Multisystem involvement was observed in 6% of anomalous fetuses.

SYSTEM	PERCENTAGE (%)
Central Nervous System (CNS)	25%
Cardiovascular System (CVS)	20%
Gastrointestinal System (GIT)	19%
Urinary System	10%
Musculoskeletal System	08%
Cleft Lip and Palate	03%
Multisystem	06%
Others	09%

Table 3: Classification as per system involved

Terminations were primarily due to congenital anomalies involving the CNS, musculoskeletal, cardiovascular, renal systems and multiple anomalies with CNS defects being the leading cause. Among the 25 cases of CNS anomalies, anencephaly was the most common anomaly.

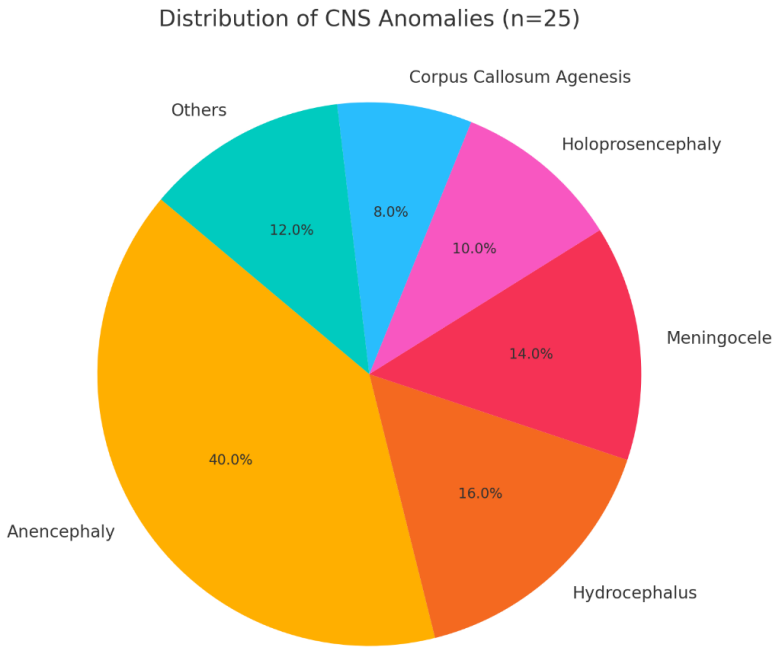

Figure 1: Distribution of CNS anomalies

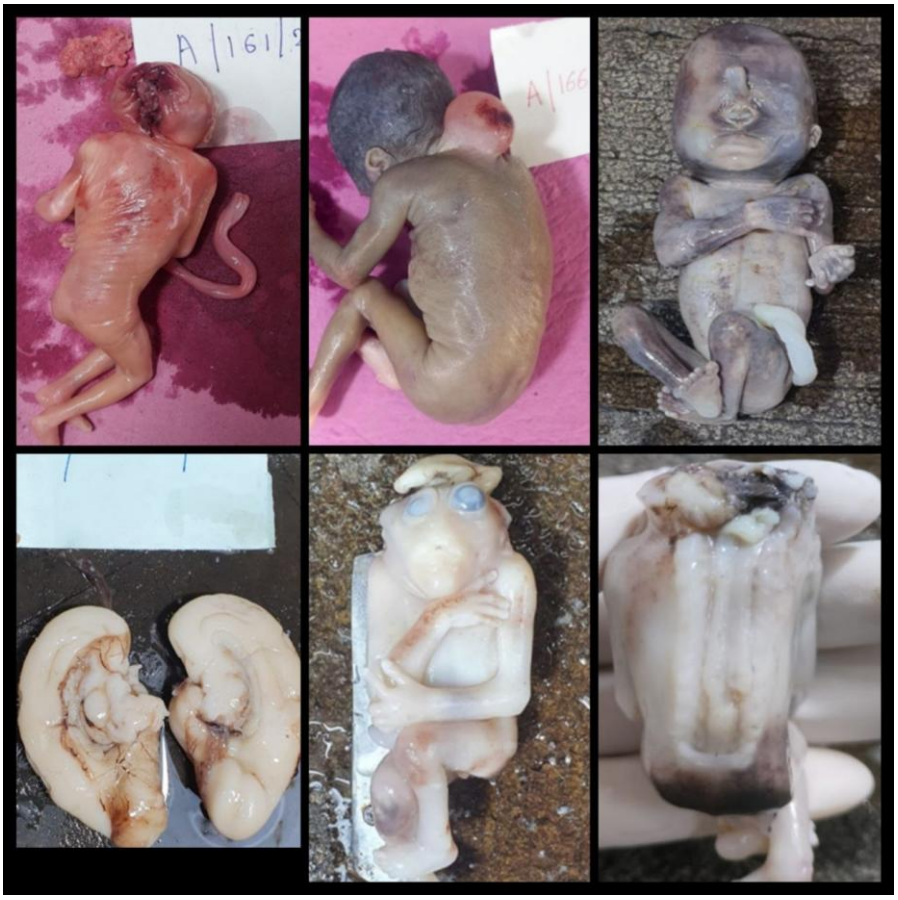

Figure 2: Various CNS anomalies- a) Anencephaly - Partial formation of cranium with rudimentary brain tissue, b) Meningocele, c) Holoproscencephaly- Facial dysmorphia with proboscis and fused eyes, d) Corpus Callosum Agenesis and e and f) Exencephaly with rachischisis

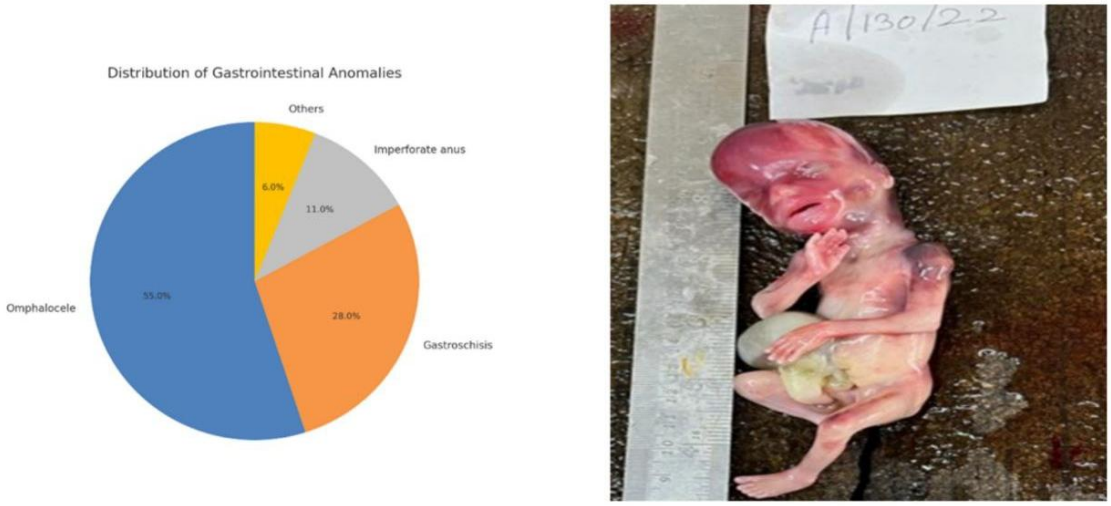

Figure 3: a) Distribution of GIT anomalies, b) Omphalocele- Abdominal organs outside the body cavity.

Omphalocele is the most common, making up 55% of cases, followed by gastroschisis at 28%.

Circulatory system showed different anomalies involving, most common being Ventral Septal Defect (VSD-62%) along with rare conditions like dextrocardia. Respiratory system anomalies included bronchogenic cysts (most common-57%), Congenital Cystic Adenomatoid Malformation (CCAM), Pulmonary hypoplasia and agenesis.

Urogenital anomalies included Horse-shoe kidney (most common-48%), Renal agenesis, Polycystic Kidney Disease (PCKD) and ambiguous genitalia (2%).

Rare cases including Potter syndrome, Ellis Van Creveld syndrome, Sirenomalia or Mermaid syndrome, Cystic Hygroma etc were reported. Potter syndrome showed classical Potter's facies, renal agenesis and pulmonary hypoplasia in case of severe oligohydramnios. In case of sirenomelia, defect in caudal region i.e. fusion of lower limbs was noted.

Figure 4- a) Sirenomelia-Mermaid tail appearance with lower limbs fusion, b) Cystic Hygroma-Fluid-filled sac at

neck.

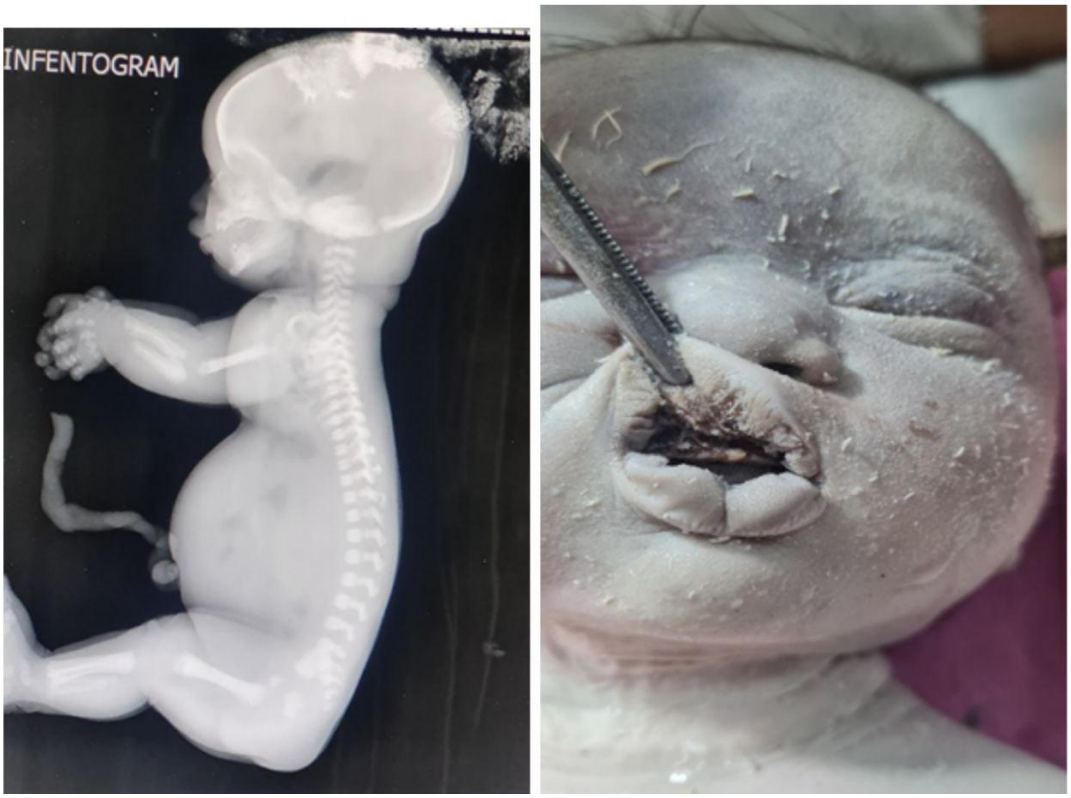

Figure 5- Ellis Van Creveld Syndrome – a) Infantogram showing small stature with short limbs, b) Presence of natal incisors in mouth.

4. DISCUSSION

Congenital effects are emerging as a major contributor to infant mortality even in developing nations where overall infant death rates have declined considerably. However, around 70% of these defects can be prevented through affordable and effective community-based genetic services. Major risk factors include early marriage, high fertility, unplanned pregnancies, poor prenatal care, malnutrition and consanguineous marriages.[7]

In this study, we observed a range of structural anomalies, with the central nervous system (CNS) and gastrointestinal (GI) tract being the most commonly affected systems.

Among CNS anomalies, anencephaly was the most prevalent, followed by hydrocephalus and meningocele aligning with global trends that highlight neural tube defects as a major concern in fetal development. In India, the incidence is reported at 4.5 per 1000 live births which is higher than in Western countries. This variation is likely due to genetic and environmental factors.[8,9] Neural tube defects like anencephaly are largely preventable with folic acid, but unplanned pregnancies in India hinder timely use. Antenatal care and food fortification with folic acid are key preventive strategies.

Study done	Prevalence rate	Findings similar to our study	Salient findings
Bhide P, Kar A[5]	184.48 per 10,000 births	Anencephaly and talipes = Most common anomalies	Only prospective study done in India
Kumar, Jogender et al.[10]	182 per 10,000 live births	Most common anomaly in CNS = anencephaly	Most common system involved - CVS

Dutta V, Chaturvedi P.[11]	48 per 10,000 live births	Most commonly seen in mid-trimester abortion	Most common system = Musculoskeletal system
Ronya R et al.[12]	21.1 per 1000 births	Male preponderance	Most common system involved = GIT
Al Dewik et al.[13]	130 per 10,000 live births	Among CVS most common anomaly= congenital heart disease	Most common system involved – CVS Risk factors analysis
Bhalerao A, Bhalerao K[14]	138 per 10,000 live births	Age group- 20 to 30 years	Most common system = Musculoskeletal system
Silest et al[15]	Prevalence rate= 5.955	Male preponderance Most common system- CNS	Linear trend analysed- Increasing prevalence

Table 4- Comparison with different studies

Among chromosomal anomalies, Down syndrome was the most common syndrome observed linked to the higher maternal age seen in this study which is observed in other studies as well. [16,17]

Both young and advanced maternal ages raise the risk of non-chromosomal anomalies. Age-specific prenatal screening is essential, especially with increasing delayed pregnancies.[18]

Our findings are consistent with prior studies conducted in similar regional settings though slight variations in frequency may be attributed to population-specific differences and sample sizes. Even though prenatal USG can detect an early malformation, a detailed autopsy examination may add up the associated anomalies seen with that specific malformation. For example, in case reported by Iqbal CW et al., antenatal USG done in second trimester showed fetal demise however etiology was not determined. External examination indicated diagnosis of amniotic band syndrome without internal examination findings. Hence, the cause of death was amniotic band syndrome (non-recurrent condition) allowing effective counseling and reassurance for the couple.[19,20] In another case, imaging showed intrauterine demise and autopsy examination revealed excessively coiled umbilical cord and growth restriction of fetus thus establishing cause of death.[20,21]

Fetal autopsy is the most definitive method for determining the cause of stillbirth. Fetal and neonatal autopsies not only help determine the cause of death but also improve clinical care, guide future pregnancy-planning, and offer closure to grieving families. It aids in evaluating diseases relevant to future pregnancies such as growth restriction, malformations, maternal diabetes or genetic disorders.[22] Identifying factors like developmental anomalies, placental pathology and maternal health conditions helps to assess risks in future pregnancies and deepens understanding of the causes behind such losses.[23,24]

In cases of congenital anomalies, genetic testing and counseling play a critical role in determining the underlying etiology, assessing the risk of recurrence, informing clinical management and supporting reproductive decision-making in future pregnancies. Wider use of genetic testing has revealed more clinically significant results aiding in improved patient care.[25]

Despite advances in genetic testing and molecular techniques, conventional autopsy remains a valuable and relevant tool for identifying the cause of death and assessing future risk.[26]

The overall distribution of anomalies in this study reflects the importance of routine antenatal care, access to diagnostic facilities and community awareness.

5. CONCLUSION

As the diagnostic gold standard, fetal autopsy pathology aids in precise anomaly detection, system-wise classification and essential genetic counseling for future pregnancies.

6. LIMITATIONS

Because of lack of financial and sample constraints, as well as a lack of infrastructure for genetic research, karyotyping of

anomalous fetuses was not feasible in this study.

The study sample included only fetuses received in department of pathology for autopsy. Hence, data was not compared with live births.

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