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Fetal autopsy: Clinicopathologic correlation in consecutive cases

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ABSTRACT

Background: Fetal and neonatal mortality serves as the most sensitive index of maternal and neonatal care. Congenital malformations remain a common cause of perinatal deaths and account for 10–15% in developing countries. Antenatal anomaly scan has become a standard element of obstetric care and their time for a fetal malformation is about 18 weeks. Fetal autopsy serves as a methodology to confirm the sonogram findings with regard to fetal anomalies. **Aims and Objectives:** The aim of our present study is to review and evaluate fetal autopsy of stillbirth/fetal losses and to confirm the malformations that were diagnosed by ultrasound. **Materials and Methods:** A cross-sectional study of a total of 10 cases received during the period of September 2023–October 2023 at the Institute of Obstetrics and Gynaecology. All the cases were spontaneous fetal losses. A standard protocol of autopsy procedure was followed which included external, internal, and histopathological examination. In fetuses with prenatally diagnosed malformations, ultrasound findings were compared with autopsy findings. **Results:** We analyzed a total of 10 fetuses, which included one chromosomal abnormality, four cardiac anomalies, one multiple anomaly, three central nervous system (CNS) anomalies, and one lethal skeletal dysplasia. The fetal autopsy confirmed the prenatal ultrasound findings in eight cases. Fetuses with CNS abnormality were not appreciated grossly due to early autolysis of the brain in the post-mortem. **Conclusion:** Prenatal imaging and screening are considered milestone development which detects fetal anomalies. However, the time-tested clinical autopsy which used to be conducted for all hospital deaths to find the cause of death was the norm of yesteryears which threw light on many unidentified causes and many syndromic associations not detected antemortem. Likewise, this case highlights the importance of autopsy correlation with prenatal imaging to highlight the same findings and to detect findings not picked up during imaging.

Key words: Fetal autopsy; Ultrasound; Congenital anomalies; Prenatal ultrasound

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INTRODUCTION

The fetal and neonatal mortality serve as the most sensitive index of maternal and neonatal care. Congenital malformations remain a common cause of perinatal deaths and account for 10–15% in developing countries.[1] The incidence of major congenital malformation is 3% and that of multiple congenital malformations is about 0.7%. In industrialized countries, malformations are the first cause of prenatal death (25–30%) and are related to elevated morbidity in the neonatal and postnatal period.[3] Antenatal anomaly scan has become a standard element of obstetric care and their time for a fetal malformation is about 18 weeks. Even though an ultrasonogram can provide a fairly accurate diagnosis, it is still necessary to examine the dead fetus for accompanying defects to confirm the diagnosis and to rule out any associated malformations. The fetal autopsy plays a vital role in the confirmation, and identification of congenital anomalies and also in the counseling of parents to prevent fetal congenital anomalies in further pregnancies.[2,12,16] Ultrasound does not have a 100% sensitivity rate for screening early anomalies,[6] nor is its sensitivity for diagnosis. The combination of antenatal sonogram and perinatal autopsy may detect the majority

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of fetal anomalies.[13] Fetal autopsy serves as a methodology to confirm the sonogram findings with regard to fetal anomalies.[5] It also helps us in finding any additional findings associated with the anomalies detected on the scan to find out any syndromic association.[15] The objective of our present study is to review and evaluate fetal autopsy of stillbirth/fetal losses and to confirm the malformations that were diagnosed by ultrasound.

Aims and objectives

The aim of our study is to review and evaluate fetal autopsy of stillbirth/fetal losses and to confirm and correlate the findings diagnosed by ultrasound.

MATERIALS AND METHODS

The present study is a case series study conducted in the Department of Pathology, Institute of Obstetrics and Gynaecology, over a period of 2 months from September 2023 to October 2023. Overall 10 cases were included in this study. Of the 10 fetuses, almost all of them were an outcome of spontaneous fetal loss. The fetal autopsy was carried out by following a standard protocol which includes fetal biometry, external examination, internal examination, and histopathogical examination. Internal organs were noted for gross abnormality. Histopathological examination of the brain, thyroid, thymus, heart, lungs, kidneys, liver, spleen, pancreas, and adrenals were carried out. The results of the autopsy and prenatal ultrasound findings were then validated.

RESULTS

Out of the 10 cases, the mean gestational age of the fetuses was 23 weeks and the men's maternal age was 28 years. After categorizing the malformation into the organ system, there were three central nervous system (CNS) abnormalities (30%), four cardiac anomalies (40%), one multi-system anomaly (10%), one chromosomal abnormality (10%), and one lethal skeletal dysplasia (10%). In our study, cardiac anomalies contribute to the major cause of death. The next common was the CNS anomalies. Chart 1 represents the categorical representation of various congenital anomalies of the fetus diagnosed on ultrasound.

In 80% of cases, fetal autopsy findings correlate with the prenatal ultrasound findings (Table 1). The cardiovascular system shows the highest agreement between prenatal ultrasound diagnosis and autopsy, while chromosomal abnormality showed the lowest disagreement between ultrasound and autopsy findings. In about 20% of cases, additional findings were obtained by fetal autopsy such as renal dysplasia, and cardiomegaly (Figure 3). In 10% of cases having CNS anomaly, fetal autopsy could not be performed because of autolytic changes. Missed anomalies may not necessarily be attributed to perinatal autopsy errors because they may also result from autolytic changes or post-mortem fetal anatomical changes.

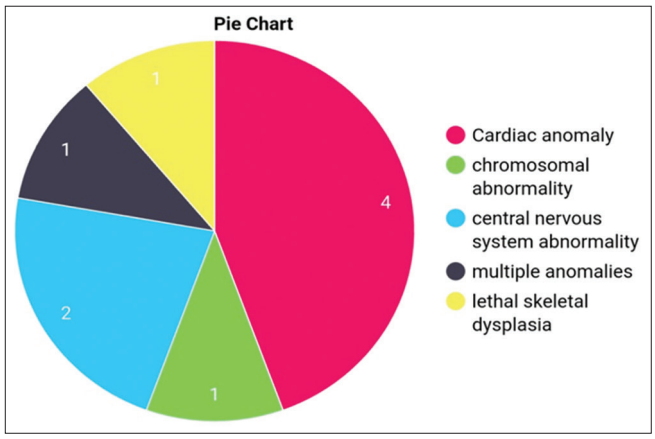

Chart 1: Categorical representation of fetal anomalies

DISCUSSION

Fetal loss is a common clinical problem and the family needs to know the cause of the loss of a fetus. The future reproductive decision of the couple depends on the cause of the fetal loss that will predict the recurrence risk and may prevent similar losses. With the advent of newer imaging techniques with higher resolution, the rate of autopsy is declining steadily. A number of studies have addressed specific organ discrepancies by comparing autopsy findings with ultrasound findings.[9] As per Vogt et al., there was complete agreement between ultrasound and post-mortem findings in 84.4% of cases, and the main diagnosis was correct in 97.8%. Only 2.2% of cases had either major autopsy findings not detected at ultrasound or ultrasound findings not confirmed by autopsy. Perinatal autopsy still remains the gold standard in detecting the cause of death and confirming the anomalies detected on ultrasound.[7] As per Rossi and Prefumo despite the high agreement between prenatal ultrasound and autopsy, the fetal examination is mandatory because, in a minority of cases, it discloses additional findings or changes the final diagnosis and genetic counseling.[4,11] Because of the increasing reluctance of parents to submit their fetus or infant to a post-mortem examination, magnetic resonance imaging has been advocated as a method to supplement or even replace autopsy.[10] However, while post-mortem MRI may be useful for gross abnormalities, it can never fully replace autopsy.[9] According to Kaasen et al., discrepancies between ultrasound and autopsy findings were observed in about 40% of the pregnancies.[8] For best outcomes, CNS fixation is essential. Complementary imaging techniques, such as MRI, may lessen the specific limitation

Table 1: The gestational age, anomaly scan findings, and fetal autopsy findings (Figures 1 and 2)

Gestational age	Anomaly scan findings	Autopsy findings
23–24 Weeks	Lethal skeletal dysplasia	Cardiomegaly, absence of cerebellar vermis, polydactyly, and atrioventricular septal defect
22 Weeks	Mild anterior cerebellar hypoplasia and prominent cisterna magna	Brain autolyzed
22 Weeks+1 day	Hypoplastic lungs, ascites, and right renal pelviectasis	Right renal pelviectasis left renal dysplasia
25 Weeks+4 days	Corpus callosum agenesis	Hypoplasia of corpus callosum
22–23 Weeks	Tetralogy of Fallot spectrum	Ventricular septal defect, overriding of the aorta, and the right ventricular hypertrophy
27 Weeks	Hypoplastic right heart syndrome	Hypoplastic right heart and doublet outlet left ventricle
24 Weeks	Large VSD with intermittent reversal of ductus	Large VSD (9–10 mm)
27 Weeks+2 days	Transposition of great arteries with VSD and pulmonary stenosis	Transposition of great arteries
17 Weeks+4 days	Trisomy 21 screen positive	Low set ears
36 weeks	Vein of Galen malformation	Vein of Galen malformation, cardiomegaly

VSD: Ventricular septal defect

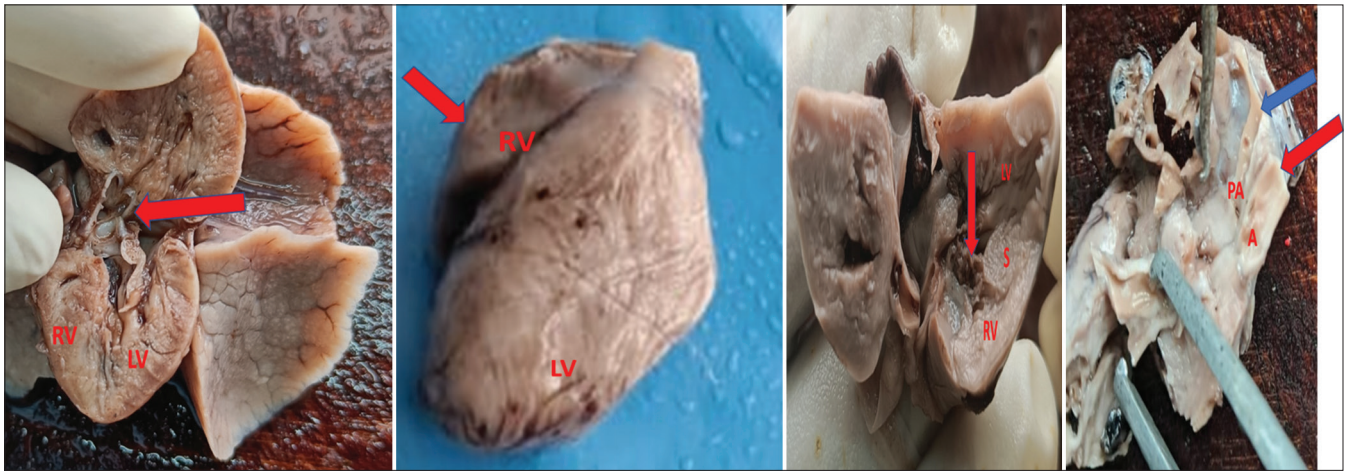

Figure 1: Top left: Tetralogy of Fallot showing overriding of the aorta, right ventricular hypertrophy. Top right: Hypoplastic right heart. Bottom left: ventricular septal defect. Bottom right: Parallelly oriented pulmonary artery and aorta arising from common left ventricle-double outlet left ventricle

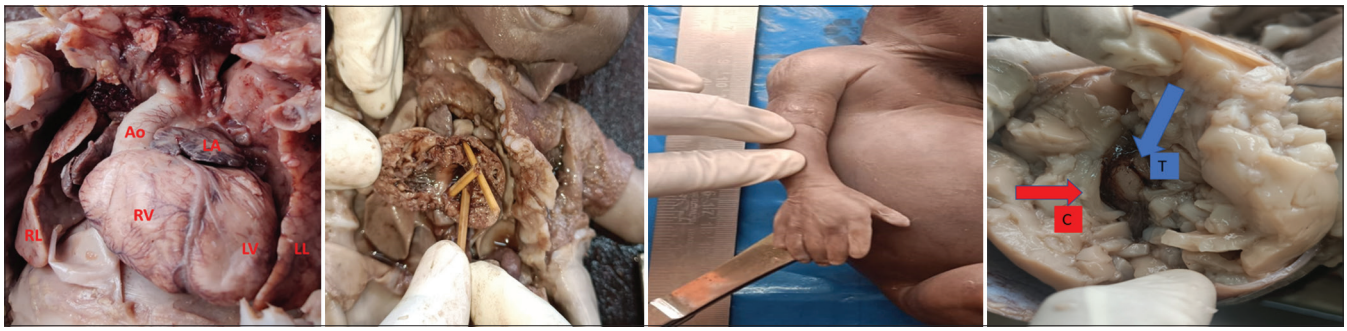

Figure 2: Top left: Transposition of great arteries. Top right: Atrioventricular septal defect. Bottom left: Polydactyly. Bottom right: hypoplasia of corpus callosum

of autopsy, that is, the detection of anomalies of CNS.[14] In addition to obstetricians, pediatricians, geneticists, and specialized perinatal pathologists have an important role in the multidisciplinary management of prenatally diagnosed fetal malformations.[17]

Hence, before a discussion of termination of pregnancy is made, it is important to have the facts accurate regarding the viability of the fetus or otherwise. It is also important that a baby who is physically and mentally challenged is not delivered to the parents and also to prevent overburdening society. Hence it is imperative that the accuracy and precision of the imaging findings is checked after the fetus is retrieved. This enables a mutual learning process that enhances the diagnostic ability of imaging and also helps in enriching the practice of perinatal clinical autopsies.

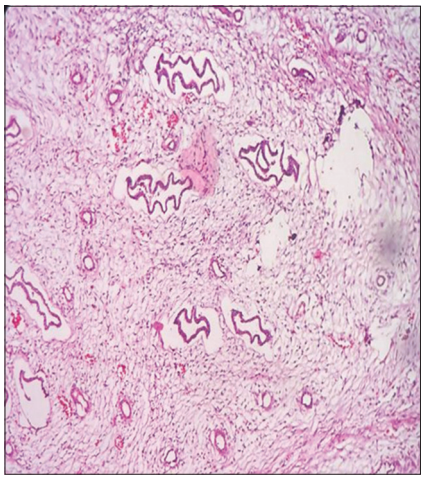

Figure 3: Renal dysplasia

This in turn will enable additional anomalies which were not evident to be found. Any syndromic association of the anomalies when found together can also be categorized, widening the knowledge about the incidence and prevalence of such conditions in our population. This article is to highlight the importance of clinical autopsy, especially perinatal autopsies in enriching the academics which subsequently will help in making informed decisions to help the patient. Do not bury the valuable information which the fetus offers without doing a clinical autopsy.

Limitations of the study

The limitations of this study is more cases need to be done to arrive at a statistically significant percentage and also imaging correlation required for all cases with more interaction between the radiologists and pathologists to arrive at a plausible consensus in how fetal autopsy will help the medical field by improving radiological diagnosis.

CONCLUSION

We believe that our data show a strong correlation between ultrasound prenatal diagnosis and autopsy results, illustrating the advantages and disadvantages of both methods. It is undeniable that autopsies play a crucial function in assisting ultrasound in reaching a precise diagnosis. Therefore, an autopsy is crucial to proper genetic counseling and correct diagnosis. Despite the advancements in imaging technology, the current study highlights the importance of fetal autopsy.

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