DOI: 10.14260/jemds/2015/328

ORIGINAL ARTICLE

FETAL AUTOPSY STUDY OVER A TWO YEAR PERIOD

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HOW TO CITE THIS ARTICLE:
Shailaja Prabhala, Padmaja Korti, Jayashankar Erukkambattu, Ramamurti Tanikella. "Fetal Autopsy Study over a Two Year Period". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 14, February 16; Page: 2263-2269, DOI: 10.14260/jemds/2015/328

ABSTRACT: INTRODUCTION: Perinatal death rate is declining in developed and developing countries and so are perinatal autopsies. In the present days due to wider availability and awareness of prenatal scanning more and more congenital malformations are picked up in earlier weeks of gestation. This helps in counselling of the couple which usually leads to an informed decision on medical termination of pregnancy. Autopsy performed on such fetuses, yields additional information in many cases. **AIMS:** The study was carried out to determine how well the prenatal ultrasound findings correlate with autopsy findings and also to determine the cause of death where ultrasound was not performed in patients who had spontaneous intrauterine death and abortion. **MATERIALS AND METHODS:** This was a prospective study carried out over a period of two years in the department of pathology at Kamineni Academy of Medical Sciences and Research Centre, Hyderabad, from January 2013 to December 2014. A total of 23 fetal autopsies were performed of which one was a twin pregnancy. Brief maternal history, prenatal ultrasound scans, relevant biochemical markers and genetic studies wherever done, were noted. **RESULTS**: There were 14 male (60.86%) and 8 female (34.78%) fetuses, and in one case (4.34%) gender could not be identified. Nineteen cases (82.60 %) were less than 28 weeks of gestation. Medical termination of pregnancy was done in 13 cases (56.52 %) whereas, 10 patients (43.47 %) had spontaneous intrauterine death of the fetus. Ultrasound scanning was done in 15 cases (65.21 %). In 13 cases (86.66 %) the ultrasound and autopsy findings were correlating whereas in two cases (13.33 %) there were findings on imaging study which could not be identified on autopsy. Ultrasound was not done in 8 cases (34.78 %) out of which 5 cases (62.5 %) showed findings on autopsy which could have led to the fetal demise. Twelve cases (52.17 %) were referral cases which had come from other hospitals. Genetic studies were done in 6 cases (26.08 %) in the form of parental karyotyping and cord blood could be tested in only one case. **CONCLUSIONS:** There is a good correlation between prenatal ultrasound scanning and autopsy findings. However, functional heart defects, and minute ventricular septal defects cannot be identified on autopsy due to the small size of the organs. At the same time autopsy can demonstrate more accurately congenital malformations and unsuspected cord abnormalities. More awareness is required on the part of treating clinicians as to the appropriate sample collection, its timely transport to the laboratory in order to facilitate genetic testing.

KEYWORDS: Fetal autopsy, congenital malformations, prenatal ultrasound, cord abnormalities.

INTRODUCTION: The term 'autopsy' derives from the ancient Greek word 'autopsia' that means 'to see for oneself' derived from (Autos- 'oneself') and (Opsis-'eye').[1] Better knowledge of unexpected fetal loss helps in parental counselling and for prevention of recurrences. Fetal autopsy can provide a clue to ascertain the cause of death in such cases.[2] Sometimes the clinicians are reluctant to request for autopsies, partly because of administrative constraints, and also due to difficulty in obtaining consent from parents or family members due to religious beliefs.[3] In India, perinatal mortality rate is

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high but perinatal autopsy is rarely performed. The ignorance about the benefits, and cost constraints, lead to reluctance for fetal autopsy.

MATERIALS AND METHODS: The present study was a prospective one, carried out over a two year period in the department of pathology at Kamineni Academy of Medical Sciences and Research Centre, Hyderabad from January 2013 to December 2014. A total of 23 consecutive fetal autopsies were performed of which one was a twin pregnancy. Brief maternal details, prenatal ultrasound scans, relevant biochemical markers and genetic studies wherever done, were correlated.

The autopsies were performed as per standard procedure and findings were recorded. The weights of the fetus and placenta, and the length of the umbilical cord were noted. After a thorough external examination, the anthropometric measurements were made which included the crown to rump, crown to heel, and foot lengths, and head, abdomen and chest circumferences. Next, a longitudinal incision was given and the thorax and abdomen were opened. Any fluid collections in the pleural and peritoneal cavities were noted.

All the visceral organs were examined to know whether they were present in appropriate location and whether any hypoplasia or absence of organs was there or otherwise. All the organs were removed en bloc and bits were given from all the organs for histopathological processing. The placenta and cord were examined thoroughly, and representative bits were taken from placenta. Two bits from the umbilical cord were taken at different levels. The fetuses were sutured and preserved in the containers.

RESULTS: As ours is a teaching hospital, in the present study 12 cases (52.17%) were referral cases which had come from neighbouring nursing homes. Eleven cases (47.82%) were from registered antenatal cases from our own institute. There were 14 male (60.86%) and 8 female (34.78%) fetuses, and in one case (4.34%) gender could not be identified. Nineteen cases (82.60 %) were less than 28 weeks of gestation. Medical termination of pregnancy was done in 13 cases (56.52 %) whereas, 10 (43.47 %) were spontaneous intrauterine deaths. Prenatal ultrasound scanning was done in 15 cases (65.21 %). In 13 cases (86.66 %) the ultrasound and autopsy findings were correlating, whereas, in two cases (13.33 %) there were findings on imaging study which could not be identified on autopsy. Ultrasound was not done in 8 cases (34.78 %) out of which 5 cases (62.5 %) showed findings on autopsy which could have led to the fetal demise. Placenta was submitted in 14 cases (63.63%) only. Thirteen cases (56.52 %) showed congenital malformations externally and/or internally, some of them having involvement of more than one system. (Table 1) (Figure 1 and 2).

Three cases (13.04%) showed placental abnormality in the form of smaller placenta and these fetuses also showed severe intrauterine growth retardation. In 5 cases (21.73%) no abnormality was found, out of which one had a ventricular septal defect and the other had a tricuspid regurgitation, both detected on ultrasound scan and confirmed by Doppler study and for both cases MTP was done. But these anomalies could not be demonstrated on autopsy due to the early gestational age, 22 weeks and 16 weeks respectively. An interesting finding was that 6 (26.08%) out of 23 umbilical cords showed abnormalities. Four cases (17.39%) showed only two vessels, one case showed two true knots and this case also had evidence of chronic placental insufficiency in the form of a smaller placenta and smaller for gestational age fetus. The other cord was extra-long and was completely twisted in its entire length with discoloration and necrosis at the umbilical site. (Figure 3)

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Cytogenetic studies on cord blood samples were advised in 12 cases (52.17%) but could be done in only one case (8.33%) due to delay in transport of the specimen to the genetics laboratory. It was reported as normal 46 XY. Most of these were referral cases. Parental karyotyping was done in six cases which were normal. Fluorescent in situ hybridization was carried out on formalin fixed paraffin blocks of products of conception in three cases for chromosome 13, 18 and 21. In all three cases aneuploidies were not found.

DISCUSSION: Congenital malformation remains a common cause of perinatal death and accounts for nearly 25-30 % in developed countries and 10-15 % in developing countries like India.[4, 5] The incidence of congenital malformations in fetal autopsies has been reported as 69 % by Kanchan et al[2] and 63 % by Puri et al.[6]. In our study the incidence of congenital malformation was 56.52 %. Open neural tube defects were the most common anomalies, 31.7 % and 33 % respectively as reported by Agarwal et al, [7] and Kanchan et al[2]. In the present study, there was not a single case of neural tube defect.

The incidence for urogenital anomalies has been reported as 16.6%,[2] and 4.2 % by Favorito et al.[8] Andola et al[9] have reported 20.45 % of renal anomalies in a study of 100 cases. In our study there were 3 cases (13.04%) having renal abnormalities in the form of nonascent and underdevelopment of one kidney, bilateral multicystic kidneys and bilateral renal agenesis in one fetus each. Gastrointestinal tract anomalies have been reported as 32%[2] while in our study, they accounted for 4 cases (17.39%).

Autopsy examination of fetuses after prenatal diagnosis of malformation is very important as it may yield additional information. In our study, autopsy examination confirmed the ultrasound findings in 86.66 % cases which is similar to the study of John et al[10] who found that there was no major disagreement between ultrasound and postmortem findings in 98% cases. But in our study, USG and Doppler studies demonstrated cardiac defects which could not be ascertained in autopsy due to the early gestational age and small size of the organs.

New findings, other than those seen in prenatal ultrasound scan may be revealed at autopsy as reported by Sunilkumar et al [11] who found additional findings in 56.3 % cases. In the present study, prenatal ultrasound scanning was done in 15 cases and autopsy was done in all cases. Additional findings on autopsy were revealed in 6 (40%) cases in the form of pancreatic cyst, anorectal agenesis in one case each and four cases of cord having two vessels only.

Pathologic abnormalities of placenta or umbilical cord lead to early intrauterine death, hence their examination becomes very important.[12] In our study, only 14 (60.86%) placentas were studied. Conditions like placental infarctions, retroplacental hematomas and chorioamnionitis are missed when placental tissue is not studied. Umbilical cord abnormalities predispose fetus to stasis-induced vascular ectasia and thrombosis leading to adverse neonatal outcome including intrauterine growth retardation and still birth.[13] In our study, 6 cases (26.08%) showed cord abnormalities.

In our study most of the products of conception had reached the genetics laboratory later than 24 hours and hence had become unsuitable for karyotyping test. More awareness on the part of referring doctors about the importance of timely transport of samples to the testing centre will overcome this problem.

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CONCLUSION: There is a good correlation between prenatal ultrasound scanning and autopsy findings. However, functional heart defects and minute ventricular septal defects may not be identified on autopsy in early gestational age due to the small size of the organs. At the same time autopsy can demonstrate more accurately congenital malformations and unsuspected cord abnormalities. More awareness is required on the part of treating clinicians regarding appropriate sample collection and timely transport to the laboratory to facilitate genetic testing which has a bearing on the counseling and risk assessment.

Findings	No. of cases	Percentage
Congenital malformations(external and /or internal)	13	56.52%
Intrauterine growth retardation	03	13.04%
Cord Abnormalities	06	26.04%
Placental abnormalities	03	13.04%
No abnormality detected	05	21.73%

Table 1: Autopsy findings in 23 cases

System involved	Number of cases	Percentage (%)
Musculoskeletal	**5**	**21.73**
Tetra-amelia	1	
Bilateral talipes deformity with syndactyly	1	
Rocker bottom feet	1	
Syndactyly in upper limb digits	1	
Few absent digits in hands	1	
Gastrointestinal	**3**	**13.04**
Anorectal agenesis with imperforate anus	1	
Large omphalomesenteric cyst with absence of distal gut	1	
Large pancreatic cyst	1	
Renal	**3**	**13.04**
Non-ascent of one kidney,	1	
Bilateral multicystic kidneys	1	
Bilateral absent kidneys with Potter's facies	1	
Central nervous system	**2**	**8.69**
Absent cranial vault bones	1	
Absent corpus callosum	1	
Cardiovascular	**1**	**4.37**
Truncus arteriosus,	1	
Respiratory	**1**	**4.37**
Congenital adenomatoid malformation of both lungs	1	

Table 2: System-wise fetal malformations on autopsy

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Cord abnormalities	No. of cases (23)	Percentage (100%)	Placental abnormalities	No. of cases (14)	Percentage (100)
Two vessels only	4	17.39%	Smaller for gestation	3	21.42%
True knots	1	4.34%			
Extra-long cord	1	4.34%			
Total	**6**	**26.08%**		**3**	**21.42%**

Table 3: Placenta and cord abnormalities

USG done (15 cases)						USG not done (8 cases)			
USG+A+		USG+A-		USG-A+		A+		A-	
No. of cases	Percentage	No. of cases	Percentage	No. of cases	Percentage	No. of cases	Percentage	No. of cases	Percentage
13	86.66	2	13.33	-	-	5	62.5	3	37.5

Table 4: Correlation of USG and autopsy findings

USG+ A+: positive ultrasound and autopsy findings
USG+A-: positive ultrasound and negative autopsy findings
USG-A+: negative ultrasound and positive autopsy findings
A+: positive autopsy findings
A-: negative autopsy findings

Figure 1: Tetra-amelia showing absence of all four limbs.

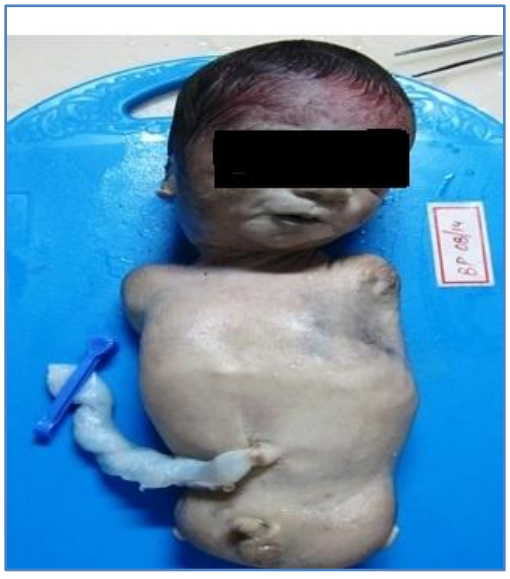

Fig. 1

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Figure 2: External surface - Bilateral congenital adenomatoid malformation of lungs showing enlarged lungs with cystic areas.

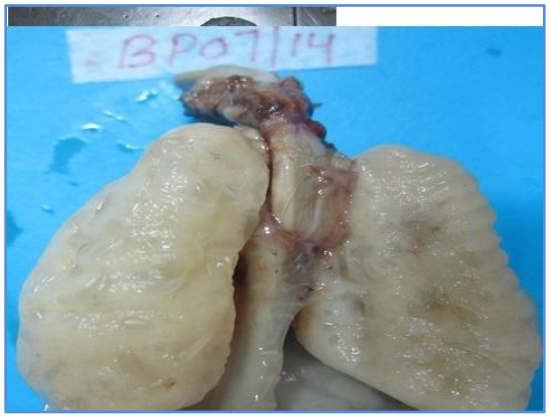

Fig. 2

Figure 3: Extra-long umbilical cord showing twisting in its entire length.

Fig. 3

ACKNOWLEDGEMENT: I wish to thank Dr. Hasan Q. Professor and Head, Dept of Genetics and Molecular Medicine, Kamineni Hospitals Ltd, Hyderabad, for providing the cytogenetic reports.

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FINANCIAL OR OTHER COMPETING INTERESTS: None

Date of Submission: 18/01/2015.
Date of Peer Review: 19/01/2015.
Date of Acceptance: 07/02/2015.
Date of Publishing: 13/02/2015.