Print ISSN: 2321-6379
Online ISSN: 2321-595X

Original Article

Comparison of Postmortem Analysis of Anomalous Fetuses with the Prenatal Ultrasonography Findings

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Abstract

Background: Congenital anomalies cause around 10–15% of perinatal deaths in India. At present, these fetal deaths are only evaluated with ultrasonography (USG) findings. With this study, we aimed to do the postmortem analysis of these anomalous fetuses and compare the findings with antenatal ultrasound findings.

Materials and Methods: A descriptive study carried out with 43 anomalous fetuses over 1½ years period in a tertiary care institute by comparing the postmortem analysis of anomalous fetuses with the prenatal USG findings.

Results: Among the 43 cases, 41 had antenatal USG taken. In 11 cases, autopsy confirmed the USG findings and autopsy showed extra findings in 16 cases. Antenatal USG was normal in 12 cases, but autopsy only identified anomalies. In two cases, autopsy detected no anomalies. Among 41 cases, 12 cases with normal antenatal USG had significant findings and 11 cases with abnormal USG had new findings after physical examination and X-ray evaluation.

Conclusion: Fetal autopsy helps in identifying many external and internal malformations which were undetected by antenatal USG. Fetal autopsy can be useful in 95% of cases – it can either confirm or add findings. Limited fetal autopsy can benefit in 56% of cases. The study shows that in majority of cases, the limited fetal autopsy itself will detect many anomalies undetected by the USG.

Key words: Anomalous fetuses, Autopsy, Ultrasonogram, X-ray

INTRODUCTION

Congenital anomalies cause around 10–15% of perinatal deaths in India.[1,2] Over the past several decades, research has dramatically improved our insight into genetic and environmental causes of many isolated birth defects, multiple congenital anomaly syndromes, and other genetic conditions. Anomalies can be malformations, deformations, dysplasia, or disruptions.[3]

When there is a fetal loss associated with anomalies, there will be an added psychological and social trauma for parents. There can be worries regarding the recurrence of these anomalies. Hence, there should be adequate evaluation of these fetuses. The risk factors and probable etiology of these anomalies should be searched for and analyzed. Thus, the evaluation of congenital anomaly starts from antenatal period, assessing the maternal factors, familial risk factors, and ultrasonogram evaluation. In the postnatal period, we can do a morphological, radiological, and histopathological evaluation with investigations for infectious, genetic, chromosomal, or metabolic causes if needed.[4]

At present, PM analysis is not being done in majority of the institutions. Decisions are made based only on the ultrasonography (USG) findings. Although some anomalies can be detected by USG, these findings can be proved right or wrong and modified with new findings after fetal autopsy. In some cases, the anomalies detected can be put together and concluded as some association, sequence, or syndrome. If necessary and feasible, confirmatory tests like karyotyping can be done for these cases. Proper genetic counseling can be done for the patients if a definite etiology is identified.

At present, these fetal deaths are only evaluated with USG findings. With this study, we aimed to do the postmortem

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www.ijss-sn.com

Month of Submission : 06-2019
Month of Peer Review : 07-2019
Month of Acceptance : 08-2019
Month of Publishing : 08-2019

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analysis of these anomalous fetuses and compare the findings with antenatal ultrasound findings.

Objectives

The objectives of this study were as follows:

1. Postmortem analysis of anomalous fetuses delivered at a tertiary care institute using
   - Relevant obstetric, medical, and family history
   - Morphological assessment
   - Radiological assessment using X-ray
   - Fetal autopsy with histopathological assessment
   - Karyotyping.
2. Asses the utility of fetal autopsy in detecting the anomalies compared to antenatal ultrasonogram
3. Asses the utility of limited fetal autopsy (physical and radiological examination).

MATERIALS AND METHODS

Study Design

This was a descriptive study.

Period of Study

The study duration was from January 2018 to June 2019.

Study Setting

Labour room, neonatal intensive care unit and pathology laboratory at a tertiary care institute.

Subjects

Definition – any anomalous fetus following termination on anomaly detection, intrauterine death, or early neonatal death (first 7 days).

Inclusion Criteria

The following criteria were included in the study:

1. Anomalous fetuses who died following the second-trimester abortion, intrauterine death, or early neonatal death (first 7 days)
2. Parents giving an informed written consent
3. Anomalies diagnosed by USG or during physical examination; minor or major; internal or external; and hydrops fetalis.

Method

The obstetricians helped in counseling the parents regarding the autopsy and informing the investigator regarding the subjects. Pathological autopsy was done at the department of pathology.

After counseling the parents and getting the consent, the importance of karyotyping was informed. If they were willing, a cardiac blood sample was taken for karyotyping. A thorough physical examination was done and looked for any anomalies. The X-ray whole body of subject was taken and the body was immersed in formalin for doing autopsy.

Observations

A total of 43 fetuses/neonates with anomalies were analyzed [Figure 1].

Of 43 cases, 19 were male, 21 were female, and 3 ambiguous [Figure 2].

Majority belonged to 21–24 gestational weeks (23.2%) [Table 1].

Twenty-five of 43 cases were small for gestation age [Figure 3]. Majority of the mothers belonged to 21–34 years [Table 2] group and were primigravida. Four mothers had

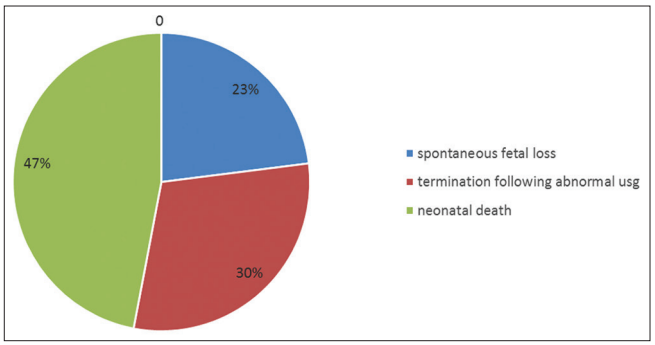

Figure 1: Subject groups

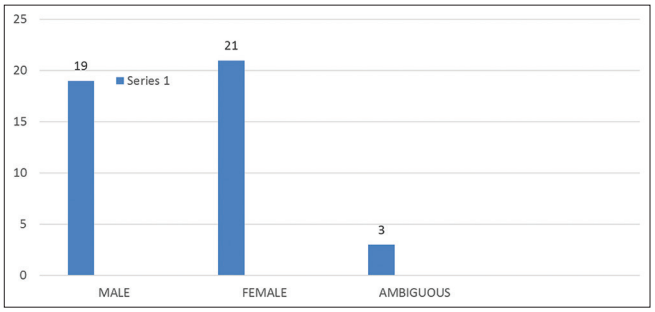

Figure 2: Sex distribution

Table 1: Gestational age

Gestational age	<20 weeks	21–24 weeks	25–28 weeks	29–32 weeks	33–36 weeks	Term
Number of cases	5	10	8	4	7	9
Percentage	11.6	23.2	18.6	9.3	16.2	20.9

Table 2: Maternal age

Maternal age	Number	Percentage
<20	6	14.3
21–34	35	83.3
>35	1	2.3

a history of stillbirths in the past and two of them were congenital anomalies [Figure 4]. Only a very few had any significant medical history. They include congenital heart disease – atrial septal defect, diabetes, hypothyroidism, and infertility treatment.

Details of ultrasonogram were available in 41 subjects. No anomalies were detected in 10 subjects. Many non-specific findings such as intrauterine growth restriction, oligohydramnios, and polyhydramnios were also taken into account [Table 3].

Among the 43 cases, 41 had antenatal USG taken. In 11 cases, autopsy confirmed the USG findings and autopsy showed extra findings in 16 cases. Antenatal USG was normal in 12 cases, but autopsy only identified anomalies. In two cases, autopsy detected no anomalies [Figure 5].

Among 41 cases, 12 cases with normal antenatal USG had significant findings and 11 cases with abnormal USG had new findings after physical examination and X-ray evaluation [Figure 6].

DISCUSSION

Congenital anomalies are associated with a major proportion of fetal losses and early neonatal deaths. Parental anxiety can be further aggravated by the association of anomalies in fetal loss. Their major concern will be of the chances of recurrence of these anomalies. Before embarking on this study, the anomalous fetuses were analyzed solely based on the findings of antenatal ultrasonogram.

We studied 43 cases with anomalies which included pregnancy terminated after detecting anomalies in antenatal ultrasonogram, stillbirths, and early neonatal deaths; the study intends to reiterate the importance of postmortem analysis of fetuses [Table 4].

Postmortem analysis included relevant history regarding family, medical illness of mother, past and present obstetric history, antenatal ultrasonogram findings, and natal and postnatal events in neonatal deaths; chromosomal analysis, physical examination, radiological evaluation [Table 5], and internal examination with histopathological evaluation. In this study, we performed the above measured in all cases except chromosomal analysis which was done in three cases due to unavailability and the cost factor of the procedure. Other investigations required were those for infections and metabolic causes.

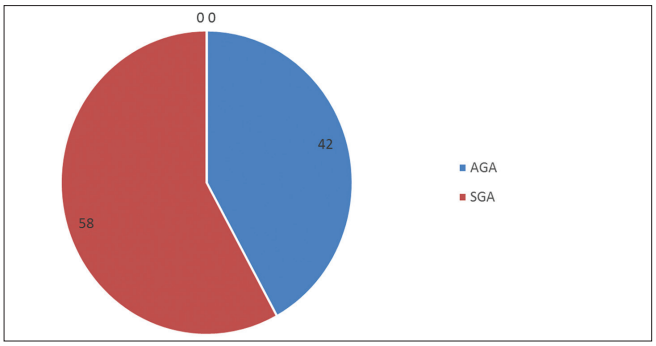

Figure 3: Weight

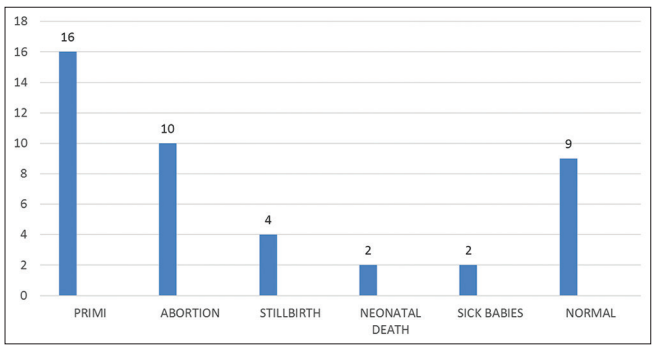

Figure 4: Obstetric history

Table 3: Anomalies found in ultrasonography

Finding	Number of cases	Percentage
No anomalies	10	24.3
IUGR	25	60.9
Oligohydramnios	7	17
Polyhydramnios	4	9.7
Hydrops fetalis	4	9.7
Renal anomalies	8	19.5
Renal agenesis	3	
Echogenic kidney	2	
Hydronephrosis	1	
Renal cyst	1	
Renal dysplasia	1	
Head, neck, and spine	16	39
Hydrocephalus	3	
Encephalocele	1	
Meningocele	1	
Anencephaly	1	
Brain malformation	1	
Arnold Chiari malformation	1	
Spina bifida	1	
Hemivertebrae	1	
Cleft palate	1	
Cystic hygroma	1	
Occipital swelling	1	
Curved spine	2	
Thoracoabdominal	12	27.9
Lung hypoplasia	2	
Cardiomegaly	2	
Endocardial cushion defect	1	
Cardiac hypoplasia	1	
Cystic mass in abdomen	1	
Thoracoomphalophagus	1	
Duodenal atresia	1	
Diaphragmatic hernia	1	
Omphalocele	2	
Limb anomaly	4	9.7

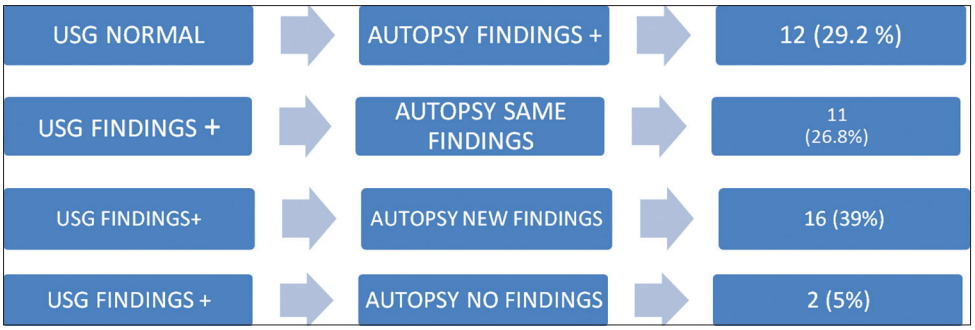

Figure 5: Correlation between ultrasonography and autopsy findings

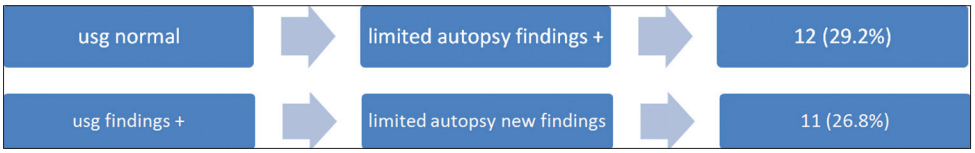

Figure 6: Correlation between ultrasonography and limited autopsy

Table 4: Physical examination finding (*n*=43) (one subject can have more than one anomaly)

Findings	Number of cases	Percentage
No anomalies	6	13.9
Hydrops fetalis	8	18.6
Head and neck	22	51.1
Anencephaly	1	
Cleft lip	3	
Cleft palate	5	
Facial dysmorphism	4	
Anophthalmia	2	
Anotia	2	
Coloboma iris	1	
Swelling neck	2	
Aplasia cutis	1	
Hemangioma	1	
Thoracoabdominal	17	39.5
Congenital heart disease	2	
Diaphragmatic hernia	1	
Meningomyelocele	1	
Thoracopagus	1	
Kyphoscoliosis	1	
Ambiguous genitalia	3	
Imperforate anus	2	
Single umbilical artery	3	
Omphalocele	2	
Gastroschisis	1	
Limb anomalies	10	23.2
Syndactyly	3	
Polydactyly	1	
Talipes equinovarus	2	
Joint dislocation	1	
Bowing of legs	1	
Absent fingers	1	
Ectrodactyly	1	

Table 5: X-ray findings (*n*=43) (one subject can have more than one finding)

Finding	Number of cases	Percentage
No skeletal anomaly	26	60.4
Thorax	9	20.9
Skull and spine	5	11.6
Limbs	6	16.2
Lungs	7	16.2
Congenital lobar emphysema	3	
Congenital cystic adenomatoid malformation	1	
Hypoplastic lungs	2	
Lung cyst	1	
Renal	7	16.2
Renal agenesis	4	
Renal cyst	1	
Infantile polycystic disease	1	
Dilated urinary bladder with megaureters	1	
Brain	4	9.3
Hydrocephalus	2	
Arnold Chiari malformation	1	
Dandy-Walker malformation	1	
Cardiac anomalies	5	11.6
Single atria and ventricle	1	
Myocardial hypertrophy	4	
Abdominal	6	13.9
Intestinal stenosis	3	
Liver cyst	1	
Diaphragmatic hernia	1	
Rudimentary liver	1	
Others	4	9.3
Spinal hemangiomas	1	
Scalp hemangioma	1	
Lymphangioma	1	
Storage disorder – liver and heart	1	
Hydrops fetalis – unknown cause	4	9.3
Internal organs	14	32.5

Unlike other similar studies, this was a prospective study. There were eight cases of non-immune hydrops fetalis, of which four had some internal malformations.

The fetal autopsy helped detecting additional findings in 39% of cases. In 29.2% of cases, antenatal USG was normal

Table 6: Efficacy of ultrasound examination – detection rate of different types of anomalies

Types of anomaly	Anomalies detected		Percentage of anomalies detected with USG
	With USG	After autopsy	
Intracranial	4	4	100
Hydrops	5	8	62.5
Limb	4	10	40
Renal	2	7	28.5
Lung	2	7	28.5
Cleft palate	1	5	20
Cleft lip	1	3	33.3
Intestinal atresia/stenosis	Nil	3	0
Cardiac	Nil	5	0

and fetal autopsy detected some anomalies. In 26.8% of cases, fetal autopsy confirmed the ultrasonogram findings. About 5% of cases had no anomalies after autopsy.

It was also found that certain ultrasonogram findings were either found absent or modified after autopsy, especially renal anomalies [Table 6]. After assessing the detection rate of different types of anomalies by USG, it was found that intracranial anomalies were detected in all cases, and cardiac and gastrointestinal anomalies were completely missed by antenatal USG.

With limited fetal autopsy, 29.2% of cases had some anomalies in spite of normal USG and 26.8% had new anomalies in addition to USG findings.

In a retrospective study done by Sankar and Phadke,[5] fetal autopsy detected additional findings in 58% of cases and confirmed the USG findings in 42% of cases. They were able to reach a final diagnosis in 59% of cases.

In another retrospective study by Phadke and Gupta, fetal autopsy provided a definite diagnosis in 72/91 (79.1) of the cases. Fetal autopsy confirmed the sonographic findings in 89 of 91 cases. About 97.8% autopsy helped in redefining the diagnosis and the risk of recurrence in 30 of 91 (33%) cases.

The similar previous studies all had confirmed the utility of fetal autopsy. Many studies have compared its utility with antenatal USG.[6-10]

The difference in the results of the above-mentioned studies from this may be due to the lack of experience and lack of infrastructure.

Fetal autopsies were done only rarely in our institution. With this study, we observed the practical difficulties of doing fetal autopsy as a routine for fetal loss. The study needs a coordinated involvement of three departments – obstetrics, pediatrics, and pathology. The participant of nursing staffs and paramedical staffs also is necessary.

Although the number of fetal losses during the study period was very high, there were many factors that made the parents unwilling for the study. They include certain religious beliefs which will not allow any intervention over body after death, emotional factors, especially in neonatal death, delay in getting the body after the postmortem analysis, and unawareness of labor room staff regarding the importance of postmortem analysis.

Limitations

The sample size was small.

We could not do complete evaluation in majority of cases.

Karyotyping was done only if the parents were interested after counseling, due to the financial factors and transplantation problems. It could be done only in three subjects, of which two samples became lysed and the other one was normal.

The recurrence risk could not be analyzed.

The ultrasonogram was done by different personnel and their can be interobserver variation.

CONCLUSION

This study done on 43 anomalous fetuses/newborns at tertiary institute shows the utility of fetal autopsy and limited fetal autopsy.

Fetal autopsy helps in identifying many external and internal malformations which were undetected by antenatal ultrasonogram.

Fetal autopsy can be useful in 95% of cases – it can either confirm or add findings.

It is possible to diagnose 53% of cases with fetal autopsy.

Limited fetal autopsy can benefit in 56% of cases – it can either confirm or add findings.

Although some of the anomalies cannot be characterized as any known disease, autopsy helps to describe the individual anomalies.

The study shows that in majority of cases, the limited fetal autopsy itself will detect many anomalies undetected by the ultrasonogram.

Hence, in cases, where the parents are not willing for autopsy, a limited autopsy can be done which includes physical examination and radiological examination.

Suggestions

Fetal autopsy should be made a hospital policy for all fetal loss, as this could have avoid the logistical constraints.

In cases, where the parents are not willing for autopsy, a limited autopsy can be done – which includes physical examination, photography, and radiological examination. Further, reading and consultation with experts later may yield the definite diagnosis.

For complete evacuation, other investigative modalities such as karyotyping, metabolic screening, and immunoassay need to be present in concerned hospital or nearby center.

Parents need a lot of counseling and motivation, especially in the setting of religious beliefs against autopsy and neonatal deaths.

The labor room staff needs to be aware of the significance and the different steps of fetal autopsy.

Proper communication between obstetricians, pediatricians, and pathologist is needed.

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How to cite this article: Murugalatha P, Krithiga M. Comparison of Postmortem Analysis of Anomalous Fetuses with the Prenatal Ultrasonography Findings. Int J Sci Stud 2019;7(5):26-31.

Source of Support: Nil, **Conflict of Interest:** None declared.