

## CORRELATION OF AUTOPSY AND SONOGRAPHY FINDINGS IN FETAL ANOMALIES

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**Types of Study:** Original Research Paper

**Conflict of Interest:** Nil

### ABSTRACT

**Background:** Ultrasonography in pregnant females is done without posing any risk to the fetus or mother. Ultrasonography is done as a scientific survey of the anatomy assessing the features of all the systems and all associated organs.

**Objectives:** The present study was conducted to compare and correlate the autopsy and ultrasonography findings in the fetus with congenital anomalies and also to detect contrasting findings.

**Methods:** In all the included fetuses, imperforate anal defects, gastrointestinal anomalies like Omphalocele, diaphragmatic hernia, club foot, cleft lip, cleft palate, palmar crease defect, and limb anomalies were assessed.

**Results:** The fetus was divided into four categories comparing ultrasonography and autopsy findings.

**Conclusion:** The present study concludes that autopsy and ultrasonography are largely influenced by dietary habits and economic status. Awareness among parents and early diagnosis of these malformations can help in timely MTP (Medical Termination of Pregnancy) and preventing these malformations in subsequent pregnancies.

**Key Words:** Autopsy, Categorization, congenital malformations, fetal anomalies, Sonography

### INTRODUCTION

Prenatal scans are usually done during 6-20 weeks of gestation and do not pose any harm to the mother or fetus. These ultrasonography examinations are termed prenatal scans and help diagnose the defective anomalies present in the embryo. This can further help the concerned gynecologist to plan timely MTP (Medical Termination of Pregnancy) and prevent these malformations in subsequent pregnancies.<sup>1</sup>

Ultrasonography is done without posing any risk to mother or fetus and is a part of an anatomic survey where external features are assessed along with the assessment of all the organs linked to each system.<sup>2</sup>

Detected fetal anomalies on ultrasonography can either be major anomalies or minor anomalies. Minor anomalies are usually not detected on the ultrasonographic examination. These minor anomalies have an incidence of nearly 14% among all the ultrasonography detected and reported scans. However, these minor anomalies don't interfere with the survival or well-being of the fetus, whereas, major anomalies can be fatal to the fetus.<sup>3</sup>

The present study was conducted to assess sex ratio and fetal anomalies conducted to compare and correlate the autopsy and ultrasonography findings in the fetus with congenital anomalies, and also to detect contrasting findings.

## **MATERIALS AND METHODS**

The present descriptive clinical study was conducted to assess sex ratio and fetal anomalies conducted to compare and correlate the autopsy and ultrasonography findings in the fetus with congenital anomalies, and also to detect contrasting findings. The study was conducted at Sridevi Institute of medical sciences, Lingapura, Tumakuru, Karnataka from June 2020 to June 2021 after obtaining clearance from the concerned Ethical committee. The study population was comprised of the subjects visiting the Outpatient department of Obstetrics and Gynecology of the Institute. After explaining the detailed study design, informed consent was taken from all the study subjects.

The study included a total of 48 antenatal ultrasonography findings from the pregnant females having embryologic anomalies seen on the scan and were following the medical termination of pregnancy or after delivery, fetal specimens were collected for autopsy. The specimens collected were including the placenta. All autopsies were conducted at the institute by experts in the field.

The inclusion criteria for the study were subjects where on ultrasonography, fetuses were found to be embryologically defective, and the same was confirmed on autopsy and the subjects who were willing to provide their ultrasound scan reports. The exclusion criteria for the study were subjects having fetal anomalies with missing ultrasound scans and the subjects who were not willing to provide their ultrasound scans.

After the final inclusion of the study subjects, detailed history was recorded followed by a physical examination of all the subjects. The demographics were recorded for all the subjects including maternal age, history of embryologic anomalies in the families, abortions, stillbirths, chemicals, radiation exposure, dietary habits, medicine during pregnancy, socioeconomic status, and gestation duration.

All the subjects were followed either till termination of pregnancy or till delivery, whichever was earlier. The fetus following delivery or expulsion was collected. Ultrasonography records were compared with the noted external appearance of the fetus. Collected placenta and fetus were obtained during 16-20 weeks of gestation and were fixed in 10% formalin. The autopsy was done for the fetus using forceps, scissors, and a knife.

The collected data were subjected to the statistical evaluation using SPSS software version 21 (Chicago, IL, USA) and one-way ANOVA and t-test for results formulation. The data were expressed in percentage and number, and mean and standard deviation. The level of significance was kept at  $p < 0.05$ .

## RESULTS

The present descriptive clinical study was conducted to assess sex ratio and fetal anomalies conducted to compare and correlate the autopsy and ultrasonography findings in the fetus with congenital anomalies, and also to detect contrasting findings. The study included a total of 48 antenatal ultrasonography findings from the pregnant females having embryologic anomalies seen on the scan and were following the medical termination of pregnancy or after delivery, fetal specimens were collected for autopsy.

Correlation of fetal autopsy, ultrasound, and prenatal findings was classified into the following categories based on the following criteria<sup>4</sup>

- **Category A** – Both findings (Ultrasound and Autopsy) were agreed.
- **Category B** – Additional data of other anomalies detected.
- **Category C** - Foetal autopsy revealed only certain Ultrasound findings.
- **Category D** - Ultrasound and Autopsy findings were mismatching.

On assessing the limb anomalies in the study subjects, it was seen that among 48 specimens assessed, skeletal dysplasia leading to shortening of upper and lower segments of both the limbs (Figure 1a) was seen in 6.25% (n=3) study subjects. Talipes Equino Varus was seen in 31.25% (n=15) of study subjects and fetuses. Omphalocele (Figure 1b) was seen in 4.16% (n=2) study subjects. It was associated with horseshoe-shaped kidney, fused toes, scoliosis, absence of right lower limb, edema, and short neck, whereas, other two specimens have only omphalocele as shown in Table 1. The incidence of cleft palate was also seen in 4.16% (n=2) of study subjects with associated poorly formed nose and short phallus in one subject (Table 1).

The results of the present study have also shown that diaphragmatic hernia was seen in 3 subjects in total. Diaphragmatic hernia on the right side was seen in 2.08% (n=1) subjects, whereas, diaphragmatic hernia affecting the left side was seen in the 4.16% (n=2) study subjects as depicted in table 2.

Concerning the presence of imperforate anus in the study subject's fetus assessed, it was seen that the presence of imperforate anus was found in 8.33% (n=4) fetuses, whereas, no incidence of the imperforate anus was found in the remaining 91.66% (n=44) study subjects (Table 2). On assessing the presence of a single palmer in the fetus of the present study, the results showed that single palmer in association with two lobbed lungs was seen in 2.08% (n=1) study subjects, whereas, absence of a single palmer was seen in 97.91% (n=47) study subjects as summarized in Table 2.

## DISCUSSION

The present descriptive clinical study was conducted to assess sex ratio and fetal anomalies conducted to compare and correlate the autopsy and ultrasonography findings in the fetus with congenital anomalies, and also to detect contrasting findings. The study included a total of 48 antenatal ultrasonography findings from the pregnant females having embryologic anomalies seen on the scan and were following the medical termination of pregnancy or after delivery, fetal specimens were collected for autopsy.

Limb anomalies in the study subjects were assessed, and it was seen that among 48 specimens assessed, skeletal dysplasia leading to shortening of upper and lower segments of both the limbs was seen in 6.25% (n=3) study subjects. Talipes Equino Varus was seen in 31.25% (n=15) of study subjects and fetuses. Omphalocele was seen in 4.16% (n=2) of study subjects. It was associated with horseshoe-shaped kidney, fused toes, scoliosis, absence of right lower limb, edema, and short neck, whereas, other two specimens have only omphalocele. The incidence of cleft palate was also seen in 4.16% (n=2) of study subjects with associated poorly formed nose and short phallus in one subject. These results were consistent with the findings of Nayab A et al<sup>5</sup> in 2010 and Valerie D et al<sup>6</sup> in 2011 where authors have reported similar incidence of limb abnormalities on fetus assessment in their respective studies.

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## **CONCLUSION**

Within its limitations, the present study concludes that autopsy and ultrasonography are largely influenced by dietary habits and economic status. Awareness among parents and early diagnosis of these malformations can help in timely MTP (Medical Termination of Pregnancy) and preventing these malformations in subsequent pregnancies. However, the present study had a few limitations including a small sample size, short monitoring time, and geographical area biases. Hence, more longitudinal studies with a larger sample size and longer monitoring period will help reach a definitive conclusion.

## **REFERENCES**

1. Antonsson P, Sundberg A, Kublickas M. Correlation between Ultrasound & Autopsy findings after 2nd Trimester terminations of Pregnancy. *J Perinat Med* 2008;36:59-69.
2. Kaasen A, Tuveng A, Heiberg AE. Correlation between Prenatal Ultrasound and autopsy findings: a study of second-trimester abortions. *Ultrasound Obstet Gynecol* 2006;28:925-933.

3. Taboo ZA. Prevalence and Risk Factors for Congenital Anomalies in Mosul City. Iraqi Postgraduate Med J 2012;11(4):458-470.
4. Kulkarni ML. Congenital malformation. Indian Paediatr 1989;26:5-9.
5. Nayab A, Irshad A, Amir HM. Congenital Anomalies: Prevalence of Congenital anomalies in 2nd Trimester of Pregnancy in Madina Teaching Hospital, Faisalabad on greyscale ultrasound. JUMDC 2010;1:1-6.
6. Valerie D, Luc L. Fetal and Perinatal Autopsy in Prenatally Diagnosed Foetal Abnormalities with Normal Karyotype. SOGC Tech Update 2011;267:1047-57.
7. Vimercati A, Grasso S, Abruzzese M, Chincoli A, de Gennaro A, Miccolis A. Correlation between Ultrasound diagnosis and autopsy findings of fetal malformations. J Prenat Med 2012;6:13-7.
8. Mohan H, Bhardwaj S, Bal A. Congenital Visceral Malformations Role of Perinatal Autopsy in diagnosis. Foetal Diag Ther. 2004;19:131-3.
9. Devi R, Tilak P, Rajangam S. Multiple Congenital anomalies an aetiological Evaluation. Bombay Hospital J 2007;2:64-9.
10. Pradhan R, Mondal S, Adhya S, Chaudhuri GR. Perinatal Autopsy: A study from India. J Indian Acad Forensic Med 2013;35:971-3.

**TABLES**

| <b>Anomalies</b>                                    | <b>%</b> | <b>n</b> |
|---|----------|----------|
| <b>Talipes Equino Varus</b>                         | 31.25    | 15       |
| <b>Short upper and lower segments of both limbs</b> | 6.25     | 3        |
| <b>Omphalocele</b>                                  | 4.16     | 2        |
| <b>Cleft palate</b>                                 | 4.16     | 2        |

**Table 1: Congenital anomalies seen in the study subjects**

| <b>Anomalies</b>            | <b>%</b> | <b>n</b> |
|-----------------------------|----------|----------|
| <b>Diaphragmatic hernia</b> |          |          |
| Right                       | 2.08     | 1        |
| Left                        | 4.16     | 2        |
| <b>Imperforate anus</b>     |          |          |
| Present                     | 8.33     | 4        |
| Absent                      | 91.66    | 44       |
| <b>Single palmer</b>        |          |          |
| Present                     | 2.08     | 1        |

|        |       |    |
|--------|-------|----|
| Absent | 97.91 | 47 |
|--------|-------|----|

**Table 2: Fetal anomalies seen in the study subjects**



**a) Skeletal dysplasia**



**b) Omphalocele**

**Figure 1: Fetal anomalies on ultrasonography**