Section: Pathology

Original Research Article

# STUDY OF VARIOUS CONGENITAL MALFORMATIONS IN FOETAL AUTOPSY AND CORRELATION WITH MATERNAL AND RADIOLOGICAL FINDINGS

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*Received* : 15/04/2023
*Received in revised form* : 20/05/2023
*Accepted* : 18/06/2023

*Keywords:*
*Congenital anomalies, CNS defects, anencephaly.*

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DOI: 10.47009/jamp.2023.5.4.45

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

*Int J Acad Med Pharm*
2023; 5 (4); 218-222

## Abstract

**Background:** Congenital malformations have become important cause of fetal and neonatal (perinatal) mortality in developed countries and would very soon be increasingly important determinants of fetal mortality in developing countries like India. The aim are to study and categorize the various types of congenital fetal anomalies, to correlate with maternal history, radiological finding's and determine its clinical significance and to study the pathological findings in fetal autopsies. **Materials and Methods:** This retrospective observational study was done in ESICMC KALABURAGI in Department of Pathology for a period of 2 years from JAN 2019 TO DEC 2021. **Result:** A total of 24 perinatal autopsies were done, of which 17 (70.8%) autopsies showed various congenital malformations with slight male preponderance (11 cases, (64.7%). They were common between 20- 24 wks of gestational age & birth weight range 350- 1000g. 12 (70.5%) were born to mothers in age group of 20-25 years and 11(64.7%) mothers were primipara. History of consanguinity was present in 6 mothers (35.3%). The most common congenital anomalies included Central Nervous System defects, anencephaly being commonest among them, followed by musculoskeletal anomalies. Multiple congenital anomalies were seen in 5 cases. Prenatal ultrasound finding was available in 15 cases. Autopsy diagnosis confirmed the prenatal ultrasound diagnosis in 12 cases (80%) and in 5 cases (33.3%) additional findings were observed. In 3 cases (20%) there was discordance with prenatal ultrasound diagnosis. **Conclusion:** Fetal autopsy help in identification of fetal loss, further assisting in parents counseling regarding risk of recurrent fetal anomalies, or a subsequent spontaneous abortion in future pregnancies.

## INTRODUCTION

Congenital means "Present since birth". Congenital malformation represents defects in morphogenesis during early fetal life,[1] which is a physical, metabolic or anatomic defect apparent before birth, at birth or detected during the first year of life.[2]

Congenital malformations are a major cause of fetal and neonatal deaths as well as disability cases worldwide,[3] accounting for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[4,5] The fetal and neonatal (perinatal) mortality serves as the most sensitive index of maternal and neonatal care.[6]

In the recent years, congenital disorders are becoming to be public health issue in developing countries, due to reduction of infections and malnutrition and relative increase of morbidity and mortality due to congenital malformations.[7]

The ultrasound is important diagnostic modality, used for detection of various developmental malformations. The first diagnostic ultrasound screening usually carried out around 11-13 weeks of pregnancy, when the thickness of nuchal translucency is measured and confirms the presence of the nasal bone.[8,9]

Anomaly scan has become a routine in all pregnancies during antenatal period and the ideal time to detect malformations is at around 18 weeks. Since some of anomalies may not be detected by routine ultrasonography and therefore must be followed by fetal autopsy examination to look for associated anomalies and to confirm the diagnosis.


ISSN (O): 2687-5365; ISSN (P): 2753-6556

This helps in diagnosing the cause of fetal loss and is therefore a prerequisite for genetic counseling.[10]
CNS anomalies are one of the most common groups of congenital malformations.[8,9]
A significant number of congenital anomalies cannot be determined by antenatal investigations like ultrasonography (USG) etc. for which autopsies are pereformed.[11] Termination of pregnancy for congenital anomalies, intrauterine fetal death, and inevitable abortion are common indications for fetal autopsy.[12]
This study was undertaken to study and correlate the clinical, radiological and pathological findings in fetal death, hence measure the utility of autopsy in final diagnosis.

**Aims and objectives**

1. To study and categorize the various types of congenital fetal anomalies.
2. To correlate with maternal history, radiological finding's and determine its clinical significance.
3. To study the pathological findings in fetal autopsies.

## MATERIALS AND METHODS

This descriptive analytical study was carried out in Department of Pathology, ESIC Medical College Kalaburagi for a period of 2 years from 1st January 2019 to 30th December 2021.
The present study included all the dead fetus sent for fetal autopsy. Demographic data, clinical history, maternal history, radiological and laboratory findings available were collected from the medical records department files from MRD. Pathological findings were retrieved from the Department of Pathology from Jan 2019 TO Dec 2021. Autopsy was carried out after obtaining proper written consent by the parents or relatives who brought the fetus and was done according to standard protocol. This included a photograph, external and internal examination, histopathological examination of all tissues including placenta. In cases terminated after prenatal diagnosis of malformations, the ultrasound diagnosis and post mortem diagnosis were compared to look for agreement.

## RESULTS

During this period, a total of 24 perinatal autopsies were done, of which 17 autopsies showed various congenital malformations comprising 70.8% of perinatal autopsies. In a total of 24 fetuses, there were 14 male and 10 female babies. On external examination of 24 fetus, 17 babies showed congenital malformation. These 17 perinatal autopsies with congenital malformations were further analysed. There was slight male preponderance (11 cases, 64.7%) observed in the present study.
Congenital malformations were common between 20- 24 wks of gestational age & birth weight range 350- 1000g. [Table 1]
The age of mothers of these babies ranged from 19 years to 32 years. Maximum number of fetuses with congenital anomalies, 12 (70.5%) were born to mothers in age group of 20-25 years and 11(64.7%) mothers were primipara. Among all the fetuses with congenital malformations, 10 cases (58.82%) were having low birth weight (ranging from 350-1000 grams) for the gestational age. History of consanguinity was present in 6 mothers (35.3%) out of all congenitally malformed cases. [Table 2]

**Table 1: Gestational age &Birth weight distribution of congenital anomalies**

| **Gestational age** | **Number of cases (17)** | **Percentage (%)** |
|---|---|---|
| 20- 24 wks | 10 | 58.8 |
| 25- 29 wks | 03 | 17.7 |
| 30- 34 wks | 01 | 5.8 |
| 35-39wks | 03 | 17.7 |
| **Birth weight (g)** | **Number of cases (17)** | **Percentage (%)** |
| 350-1000 | 11 | 64.7 |
| 1001-2000 | 03 | 17.7 |
| 2001-3000 | 02 | 11.8 |
| 3001-4000 | 01 | 5.8 |

**Table 2: Maternal Factors seen in malformed fetuses& neonates**

| **Character** | **Number (out of 17)** | **Percentage (%)** |
|---|---|---|
| Maternal age | | |
| <20yrs | 00 | 0 |
| 20-25 yrs | 12 | 70.6 |
| 25-30yrs | 04 | 23.6 |
| >30yrs | 01 | 5.8 |
| Parity | | |
| Primipara | 11 | 64.7 |
| Multipara | 06 | 35.3 |
| Consanguinity | | |
| Yes | 06 | 35.3 |
| No | 11 | 64.7 |


ISSN (O): 2687-5365; ISSN (P): 2753-6556

**Table 3: System wise distribution of Congenital Malformations**

| System involved | Associated anomalies | Number of cases (n=17) |
|---|---|---|
| Central Nervous system defects | | TOTAL-12 |
| Spina bifida with lumbar meningocele | | 01 |
| Spina Bifida with single umbilical artery | Single umbilical artery | 01 |
| Spina bifida, hydrocephalus | | 01 |
| Anencephaly | | 03 |
| Anencephaly with acrania | | 01 |
| Hydrocephalus due to aqueductal stenosis | | 01 |
| Dandy walker syndrome | | 01 |
| Meningomyelocele | Hydrocephalus | 01 |
| Scalp hematoma | Sinusoidal dilatation | 01 |
| Cerebellar vermis agenesis | Diaphragmatic hernia | 01 |
| 2.Musculoskeletal system | | TOTAL-03 |
| Polydactyl, short ribs | Increased nuchal thickness | 01 |
| Polydactyl | Increased nuchal thickness | 01 |
| Diaphragmatic hernia | Cerebellar vermis agenesis | 01 |
| 3.Urinary system | | TOTAL-01 |
| Polycystic kidney | | 01 |
| 4.Respiratory system | | TOTAL-01 |
| Meconium aspiration with partial atelectasis | | 01 |
| 5.Multiple Congenital Anomalies | Cerebellar vermis agenesis, left sided diaphragmatic hernia | 01 |
| 6.Syndromes | | TOTAL-2 |
| Dandy walker syndrome | Hydrocephalus | 01 |
| Hydrops fetalis | Hepatomegaly, splenomegaly | 01 |

**Table 4: External, Internal & Multisystem congenital anomalies.**

| **External congenital anomalies- TOTAL CASES- (07)** | | | |
|---|---|---|---|
| **System affected** | **Type of anomaly** | **No** | **Total (17)** |
| CNS | Spina bifida with lumbar meningocele | 1 | 1 |
| | Anencephaly | 3 | 3 |
| | Anencephaly with acrania | 1 | 1 |
| Musculoskeletal | Polydactyly, short ribs | 1 | 1 |
| | Polydactyly | 1 | 1 |
| Internal congenital anomalies- TOTAL CASES- (05) | | | |
| CNS | Hydrocephalus due to aqueductal stenosis | 01 | 1 |
| | Scalp hematoma, sinusoidal dilatation | 01 | 1 |
| Thorax | Cerebellar vermis agenesis Congenital diaphragmatic hernia | 01 | 1 |
| | Meconium aspiration with partial atelectasis | 01 | 1 |
| Renal | Multicystic kidney disease | 01 | 1 |
| Both internal and external anomalies- TOTAL CASES- (05) | | | |
| CNS | Dandy walker syndrome | 01 | 1 |
| | Hydrops fetalis | 01 | 1 |
| | Meningomyelocele with hydrocephalus | 01 | 1 |
| | Spina bifida with single umbilical artery | 01 | 1 |
| | Spina bifida with hydrocephalus | 01 | 1 |

**Table 5: Correlation of Antenatal USG findings with Autopsy findings**

| Correlation of USG findings and autopsy findings | Cases (15) | Percentage (%) |
|---|---|---|
| USG findings confirmed on autopsy<br>• No change in diagnosis<br>• Additional findings noted in the autopsy | 12<br>12<br>05 | 80<br>80<br>33.3 |
| Changes in Diagnosis<br>USG Diagnosis Autopsy Diagnosis<br>1.Polyhydroamnios Diaphragmatic hernia, left sided<br>2. Hydrops fetalis Hepatomegaly, Splenomegaly<br>3.Oligohydromnios Polycystic kidney disease | 03 | 20 |
| Total | 15 | 100 |


ISSN (O): 2687-5365; ISSN (P): 2753-6556

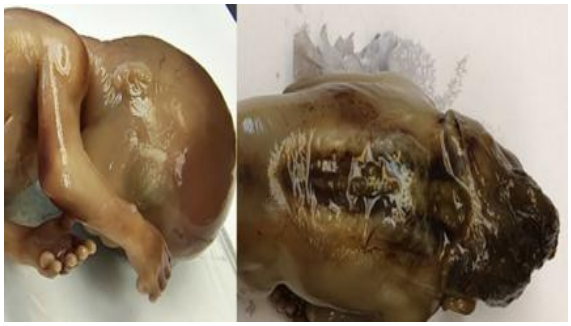

**Figure 1: a) Polydactyly. b) Anencephaly with acrania**

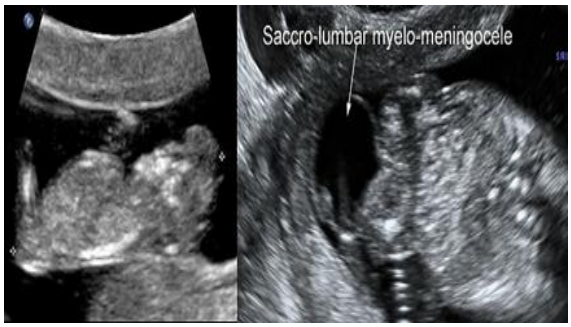


**Figure 2: Ultrasonography**

The most common congenital anomalies included Central Nervous System defects, anencephaly being commonest among them, followed by musculoskeletal anomalies [Figure 1]. Multiple congenital anomalies were observed in 5 cases. [Table 3]

On examination of 17 fetus, 7 cases showed external congenital anomalies, 5 babies showed only internal congenital anomalies. Multiple system involvement was seen in 5 cases showing both external and internal anomaly. [Table 4 & Figure 1]

Prenatal ultrasound finding was available in 15 cases. Autopsy diagnosis confirmed the prenatal ultrasound diagnosis in 12 cases and in 05 cases additional findings were observed. In 3 cases there was discordance with prenatal ultrasound diagnosis. [Table 5 & Figure 2]

## DISCUSSION

Congenital malformations are major cause of fetal death and autopsy significantly helps in diagnosis of Intrauterine fetal death.[13] The family needs to know the cause of fetus loss. The future reproductive decision of the couple depends on its cause, that will predict the recurrence risk and may prevent similar losses.[14] Detecting anomalies and malformations prenatally by various investigations like ultrasonography, chromosomal analysis, chromosomal analysis, fetal autopsy and placental examination helps in reducing the risk of recurrence which could be otherwise up to 25%.[10,15]

Few congenital malformations are preventable by following certain simple precautions such as intake of folic acid during early pregnancy i.e., the 3rd - 8th weeks of gestation. In this period along with genetic factors, various other factors like environmental, teratogenic and infectious agents play important role for the causation of malformations The presence of congenital malformation in baby has immense emotional effect on mother mental health.[2]

In the present study of 24 perinatal autopsies, 17 (70.8%) cases of congenital anomalies were encountered with similar incidence seen in Kapoor et al 2013 (69%),[16] and Venkataswamy et al 2018 (63%).[8,17]

Fetal Factors: On comparing the sex ratio, the congenital malformations were slightly more in males (64.7%) with male to female ratio being 1.8:1. Kale et al,[2] Kapoor et al 2013 (69%)16, noted higher incidence in their study. Pattanaik T et al,[18] have found the incidence to be more in males (56%) with ratio of 1.37:1. Same incidence in males and females was mentioned in some studies.

Gestational age of the fetuses with congenital malformation ranged from 20-24 weeks (58.8%) similar to study done by Pushpa B et al.[3] This may help in therapeutic termination of pregnancy after detecting malformation on ultrasonography. The association of low birth weight for gestational age and malformations has been well documented.[19] Fetuses with congenital malformations had the weight in the range of 350-1000 grams (low for gestational age) in the present study similar to study done by kale et al.[2]

Maternal Factors: Maternal age is considered as one of the important factors in incidence of congenital anomalies.[20] Maximum number of fetuses 12 (70.5%) were born to mothers in age group of 20-25 years. This finding was found similar to the study done by Subhashini et al,[21] and Kapoor et al.[16]

Bad Obstetric history, with one or more than one abortion; was present in 5 mothers (29.4%). Similar results were noted by Kale et al (24.56%),[2] Subhashini et al with 33.4%.[21]

Among the malformed fetuses or neonates, 64.7% were born to primigravida. Similar finding was seen by Kale et al (50.87%),[2] Kapoor K et al (50%),[16] Subhashini et al (40.1%),[21] and Parmar A et al (42%).[19] Consanguinity has been described as an important factor contributing to increased congenital malformations. In this study, the consanguinity was noted in 35.3% cases, while study done by Pattanaik T et al, 2016 showed less incidence (6%).[18]

The most common defects were of Central Nervous System, seen in 12 cases (70.6%) followed by musculoskeletal which correlates with the study of Kale et al, Andola US et al,[23] Potekar et al,[24] and Tomatir et al.[25] The most common defect was Anencephaly with 4 cases (25%), which was accurately diagnosed with Ultrasonography.[2]

Antenatal sonography is developed few decades back; however, it continues to lag behind a complete fetal autopsy in accurately diagnosing the cause of fetal death. Only few studies have shown comparison of antenatal ultrasonographic findings & autopsy findings. In the present study, autopsy diagnosis confirmed ultrasonographic findings in 12 cases (80%), among these in 5 cases (41.6%) it


ISSN (O): 2687-5365; ISSN (P): 2753-6556

provided additional information, whereas primary diagnosis given by ultrasonography was changed in 3 cases (25%). The findings in the present study are similar to those of Kale et al,[2] and Sankar and Phadake et al.[22]

The presence of multiple anomalies in single fetus could be attributed to same developmental period of embryogenesis of different systems. In this study, 5 out of 17 (29.40%) cases showed multiple malformations occurring in single fetus. Variety of syndromes have been identified in the study like Dandy walker syndrome, Hydrops fetalis. Antenatal detection of such malformations is important, in planning the management, and to reduce the recurrence and hence achieve better outcome.

## CONCLUSION

The present study show that a good fetal autopsy is useful in detecting and confirming congenital malformations. Despite advances in imaging such as antenatal ultrasonography and serology, fetal autopsy continues to play an important role in diagnosing congenital malformations.

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ISSN (O): 2687-5365; ISSN (P): 2753-6556