

**“ STUDY OF CORRELATION BETWEEN PRENATAL ULTRASOUND AND  
MORPHOLOGICAL FINDINGS IN FETAL AUTOPSY AT TERTIARY CARE  
CENTRE. ”**

**By**

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**DISSERTATION SUBMITTED TO**



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**IN PATHOLOGY**

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## **LIST OF ABBREVIATIONS**

ADCA: Anterior descending coronary artery

CHD: Congenital heart disease

CTEV: Congenital talipes equino varus/ valgus

IUD: Intrauterine death

IVC: Inferior vena cava

LA: Left atrium

LV: Left ventricle

MV: Mitral valve

PDCA: Posterior descending coronary artery

PV: Pulmonary veins

RA: Right atrium

RV: Right ventricle

SVC: Superior vena cava

TV: Tricuspid valve

USG: Ultrasonography

CNS: Central nervous system

SUA: Single umbilical artery

HLHS: Hypoplastic left heart syndrome

GIT: Gastrointestinal tract

IUGR: Intrauterine growth restriction

AVSD: Atrioventricular septal defect

VSD: Ventricular septal defect

PAS: Periodic acid schiff

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## **ABSTRACT:**

**TITLE:** Study of correlation between prenatal ultrasound and morphological findings in fetal autopsy at a tertiary care center.

**BACKGROUND:** Congenital anomalies are one of the major causes of fetal loss and accounts for 8%-15% of perinatal deaths in India. Amongst these, 3% neonates suffer from major congenital malformations and 0.7% to 1% with multiple system congenital malformations. When a serious fetal anomaly is suspected in the scans, obstetrician may suggest termination of pregnancy in order to avoid the heavy emotional and socioeconomic burden on both the fetus and parents or in cases where it is implied that the anomaly is almost certainly lethal. Fetal autopsy defines the examination of external and internal body parts after the demise of a fetus due to an elective termination of pregnancy or congenital anomalies and is considered as gold standard to evaluate the efficiency of ultrasound findings, along with confirming these findings to arrive to an accurate diagnosis.

**AIMS:** To compare prenatal ultrasonographic findings with fetal autopsy and to study the role of fetal autopsy in making the diagnosis of various fetal anomalies in cases of spontaneous miscarriage and medically terminated fetuses for structural anomalies.

**MATERIALS AND METHODS:** Hospital-based cross-sectional study with sample size of 150 fetuses. The study was conducted from April 2023 to December 2024. For all cases, consent was taken from parents and standard autopsy protocol were followed and histopathological examination was done. Photodocumentation of internal and external anomalies for majority of cases was maintained.

**RESULTS:**

A total of 150 fetuses were included. On comparing ultrasound reports and autopsy findings there was complete concordance in 54.6% cases (82/150), additional findings on autopsy in 39.4% cases (59/150) cases and could not confirm the ultrasound findings in 6% cases (09/150). The most common system involved was central nervous system comprising of 34 out of 150 cases (22.6%), followed by skeletal system with 16 cases (10.6%) and then cardiovascular system with 13 cases (8.6%). We also came across 23 syndromes, for which complete concordance between ultrasound and autopsy was seen in 04 cases out of 23 (17%), 07 cases out of 23 (31%) were suspected with syndrome on ultrasound and confirmed through autopsy with minor additional findings that did not lead to change in final diagnosis and 12 cases(52%) were classified into syndromes solely on the basis of autopsy.

**CONCLUSION:**

Study confirms the need for fetal autopsy after fetal loss to identify and confirm the underlying cause, which further aids in genetic counselling of the couple. Fetal autopsy plays an important role in analysing the recurrence risk thus helping in better planning for future conception plans of the parents.

Key words: Fetal autopsy, malformation, anomaly scan



## **INTRODUCTION:**

- Prenatal development of fetus is divided into three periods: 1)ovular period or germinal period-first two weeks following ovulation, 2) embryonic period- from third week of ovulation upto ten weeks of gestation (8 weeks post conception) and 3) Fetal period- from eighth week of conception until the time of delivery (generally 38 weeks of conception). The perinatal period is defined as the period starting from the conception of the fetus upto a year after giving birth <sup>(1, 2)</sup> .
- Fetal autopsy defines the examination of external and internal body parts after the demise of a fetus due to an elective termination of pregnancy or congenital anomalies. It often involves extraction of small tissue fragments for further histopathological examination. Perinatal autopsy helps in providing important details in regards to the underlying cause of fetal death to the parents and clinicians. In cases of malformations, autopsy may confirm, modify or debar the prenatal diagnosis. In cases of miscarriage, termination of pregnancy due to fetal issues or stillbirth, autopsy details provide valuable insights. These findings support parents and healthcare professionals in planning for future pregnancies and understanding potential risks, offering hope and guidance for the journey ahead <sup>(3)</sup> .
- Congenital anomalies are one of the major causes of fetal loss and accounts for 8%-15% of perinatal deaths in India. Amongst these, 3% neonates suffer from major congenital malformations and 0.7% to 1% with multiple system congenital malformations <sup>(4)</sup> . The recurrence risk generally depends on the underlying cause, for instance, single congenital malformation has a probability of 1% to 25% recurrence depending on the genetic component and other contributing environmental factors <sup>(5)</sup> .

- Fetal autopsy is considered as gold standard to evaluate the efficiency of ultrasound findings, along with confirming these findings to arrive to an accurate diagnosis. The ideal time for scan for fetal malformations is about 18 weeks and although ultrasound is reasonably precise in making a diagnosis, fetal autopsy is recommended as a supportive investigation of the terminated fetus to acquire additional useful information as to underlying case of the demise. Autopsy is also helpful in attaining tissues and DNA samples for further testing that is required for karyotyping and other additional investigations <sup>(5)</sup> .
- When a serious fetal anomaly is suspected in the scans , obstetrician may suggest termination of pregnancy in order to avoid the heavy emotional and socioeconomic burden on both the fetus and parents or in cases where it is implied that the anomaly is almost certainly lethal <sup>(6)</sup> .
- Even though the technical advancements in imaging modalities have improved in leaps and bound it should be noted that many a times these modalities can either misread and altogether fail to read certain findings. These slips can be due to several factors including position of the fetus, maternal body fat being high, previous history of fetal anomalies and oligohydrominos. Thereby, autopsy can provide a comprehensive and visual explanation for the fetal loss, often either confirming the ultrasound findings or providing additional information on the missed deformities, thus in turn providing a sense of relief to the parents as well as obstetrician about their decision to terminate <sup>(5, 6)</sup> .

### **AIMS AND OBJECTIVES:**

- To compare prenatal ultrasonographic findings with those found in fetal autopsy.
- To study the role of fetal autopsy in making the diagnosis of various fetal anomalies in cases of spontaneous miscarriage and medically terminated fetuses for structural anomalies.

## **REVIEW OF LITERATURE**

- Autopsy is a greek word derived from autos meaning self and optos meaning seen. It was termed in early 17<sup>th</sup> century in the sense of personal examination and later came to be used only in medical terms to denote postmortem examination <sup>(7)</sup>.
- Greek physicians about three millenia ago started developing interest to identify the cause of death by performing autopsy. This lead to gross dissection of human body becoming an integral part of medical education starting from regions like paris, padua and parma <sup>(7)</sup>.
- The major advances in this field came about in the 19<sup>th</sup> century due to two renowned pathologists, Karl Von Rokitansky and Rudolph Ludwig Karl Virchow <sup>(7)</sup>. Rokitansky majorly focused on developing a meticulous way of dissection for the purpose of demonstrating gross autopsy findings to achieve a clinocopathological correlation. To master the procedure Rokitansky performed 30,000 and supervised 70,000 autopsies. Similarly, Virchow agreed on the importance of gross examination, but was also engaged in the significance of microscopic findings, as he strongly believed that the disease began at a cellular level. Both scientists were also responsible in devising two principal techniques of gross examination that is in situ method of organ examination and organ block method respectively <sup>(7, 8)</sup>.
- Perinatal autopsies are different from adult autopsies in many ways , for instance, instruments used have to be smaller to account for the intricate and delicate organs to be grossed, the external examination that requires a detailed account of all morphological features ranging from skeletal dysplasia, cleft lip, abnormal head size and shape, stenosis and atresia of anus, low set ears and other features and finally intensive histological

sectioning of lungs, liver, kidney, thymus and brain to analyze the cellular development and related conditions <sup>(5)</sup> .

Types of birth defects<sup>(9)</sup>:

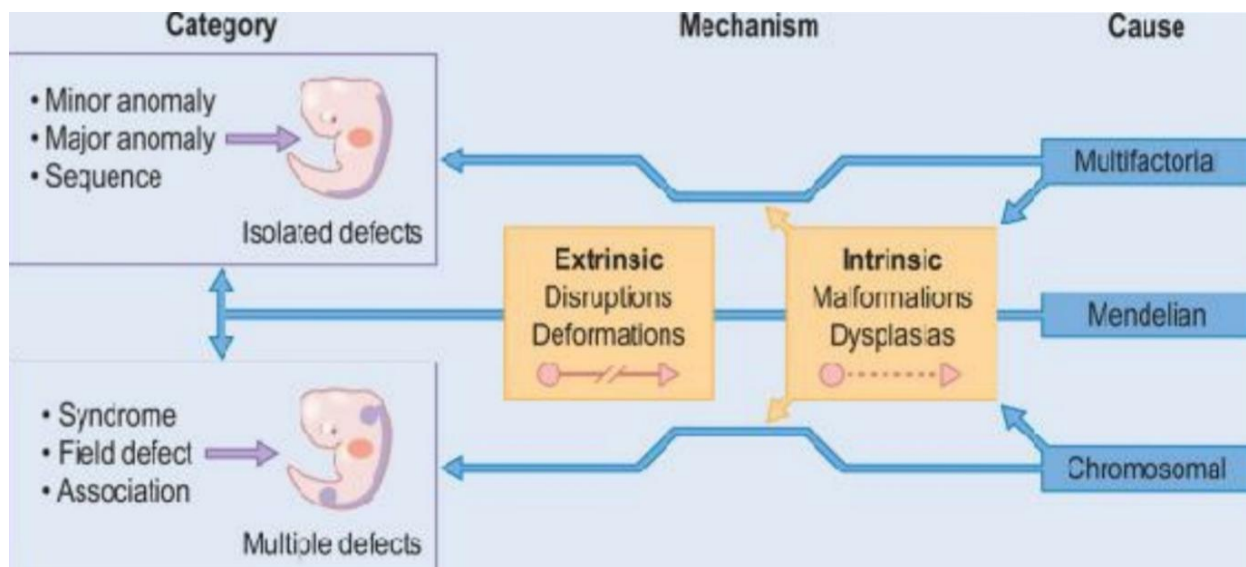


Figure 1: Categories of birth defects <sup>(9)</sup>

1) Isolated defects: Affect single body region and are usually considered due to multifactorial determination.

a) Anomaly: Alteration from the presumed type of structure, form and/ or function that may be interpreted to be abnormal.

Major anomaly: Those with cosmetic and/or surgical consequences (deformed wrist, limb defect, neurofibromas).

Minor anomaly: Have diagnostic importance but display very little impact on individual well being (anteverted nares, epicanthal folds, single palmar crease).

- b) Malformation: Morphological defect of an organ or a region of a considerable size of body due to inherently flawed ( chromosomal abnormality, Mendelian mutation, genetic predisposition) developmental process.
  - c) Dysplasia: Unusual cell arrangement into tissue and its morphological result, that is, abnormalities in histogenesis.
  - d) Disruption: Morphological defect of large region of body or a specific organ due to extrinsic breakdown or hindrance in normal developmental process. (amniotic band wrapping around developing limbs).
  - e) Deformation: Abnormality in a body part's shape, form, or location brought on by mechanical forces. (radial club hands in radial aplasia).
- 2) Sequence: Pattern of multiple anomalies derived from single known or prior anomaly or mechanical factor. It represents a course of primary or secondary occurrences that follow after a single primary disruption or abnormality.
- 3) Multiple defects: More likely due to chromosomal or mendelian inheritance.
- a) Syndrome: Several defects that are believed to be pathogenetically connected but do not constitute a sequence. The condition could eventually turn into a sequence.
  - b) Association: Nonrandom occurrence of multiple congenital anomalies in two or more individuals not known to be polytopic defect, sequence or syndrome. (VATER association, MURCS association, CHARGE association, tracheal- esophageal association, otocephaly association).

- c) Developmental field defects: It is caused by result of disturbed development of a morphogenic field or of a part. It is important to note that most field defects carry very low genetic risk if present in isolation.
- 4) Qualitative terms:
- a) Hyperplasia or hypoplasia: Refers to the overdevelopment or underdevelopment of an organ, or tissue as a result of either an increase or decrease in the number of cells.
  - b) Hypotrophy or hypertrophy: Decrease or increase in the size of cells, tissue or organ respectively.
  - c) Agenesis: Represents the loss of a part due to absence of primordium (rudiment of organ).
  - d) Aplasia: Absence of a part of body due to lack of primordium to develop further.
  - e) Atrophy: Indicates decrease in cell size and/ or cell number of a normally developed mass of tissue or organ.
- Some important definitions <sup>(10)</sup>:
    - 1) Embryo- from the moment of conception until the conclusion of organogenesis (about eight weeks after conception), during which the blastocyst and inner cell mass develop.
    - 2) Fetus- from the end of embryo stage (8 weeks post conception) till delivery.
    - 3) Intrauterine fetal death- Death prior to the complete expulsion or extraction from its mother of a product of conception, irrespective of duration of pregnancy; the death is indicated by the fact that after such separation the fetus does not breathe or

show any other evidence of life, such as beating of the heart, pulsation of umbilical cord or definite movement of voluntary muscles without specification of duration of pregnancy.

4) Still birth- Death of fetus that has reached a birth weight of 500 grams or birth weight is unavailable with gestational age of equal to or more than 22 weeks or crown to heel length of 25 cm or more.

**Tulip classification:** On the basis of underlying cause and mechanism of perinatal mortality<sup>(11)</sup>.

Cause of death is defined as initial pathophysiological entity initiating the chain of events that has irreversibly lead to death.

Mechanism of death is defined as organ failure that is not compatible with life, initiated by the cause of death.



**Table 1: Tulip classification of perinatal mortality, causes of death <sup>(11)</sup>**

<b>Cause of death</b>	<b>subclassification</b>	
Congenital anomaly	Chromosomal defect	Numerical Structural Microdeletion/uniparental disomy
	syndrome	Monogenic Others
	Central nervous system	
	Heart and circulatory system	
	Respiratory system	
	Digestive system	
	Urogenital system	
	Musculoskeletal system	
	Endocrine/ metabolic system	
	Neoplasm	
	other	Single organ Multiple organ
Placenta	Placental bed pathology	
	Placental pathology	Development Parenchyma location
	Umbilical cord compression	

	NOS	
Prematurity/immaturity	PRROM	
	Pre-term labor	
	Cervical dysfunction	
	Iatrogenic	
	NOS	
Infection	Transplacental	
	Ascending	
	Neonatal	
	NOS	
Other	Fetal hydrops of unknown origin	
	Maternal disease	
	trauma	Maternal Fetal
	Out of ordinary	
unknown	Despite thorough investigation	
	Important information missing	

### **Tulip classification of perinatal mortality, mechanism of death<sup>(11)</sup>**

Cardiocirculatory insufficiency
Multi-organ failure
Cerebral insufficiency
Respiratory insufficiency
Placental insufficiency
unknown

- In 2006 Kaasen A et al.,<sup>(12)</sup> set out to evaluate the discrepancies between ultrasonographic and autopsy findings for second trimester abortions. They covered 274 cases by performing autopsies. In 58.4% (160 out of 274) cases there was a full agreement. Additional findings detected by autopsy that were initially not found in ultrasound was seen in 31.4 % of cases. Of a total of 64 malformations , 30 were considered as detectable. In one of the cases postmortem radiology examination was used to confirm the ultrasound diagnosis. Discrepancy between ultrasound and postmortem findings were observed in 40% of cases, thus further stressing on the need for autopsy.
- Sankar.V.H et al.,<sup>(13)</sup> from India studied 206 fetuses in 2006 to assess how useful fetal autopsy is for making a final clinical diagnosis and comparing the autopsy findings to estimate how beneficial fetal autopsy can be in terminated fetuses subsequent to the diagnosis of malformation. Of these 206, 138 were terminated post detection of an anomaly in ultrasound and 68 were spontaneous fetal losses. For prenatally diagnosed defects in

fetuses, ultrasonography results and autopsy results were compared. In 59 cases, the autopsy confirmed the ultrasound results, and in all but two cases, the fetal autopsy gave a definitive diagnosis. Furthermore, in 77 cases, autopsy revealed new information and lead to significant change in recurrence risk in 24 cases.

- In a study conducted by Amini.H et al.,<sup>(14)</sup> in the year 2006 on 328 fetuses with the objective to compare antenatal diagnoses with autopsy findings in fetal anomalies and to analyze the quality of antenatal fetal diagnosis. They observed that in 91.2% cases ultrasound either exactly matched or were very similar to the autopsy findings. Although, in 23 cases that is about 7% autopsy could not confirm the ultrasound findings but the findings of both were similar in severity. In 1.8% of terminations the autopsy findings were found to be less severe than ultrasound findings. Fetal autopsy also lead to further diagnostic information in 47% cases disclosing existence of a syndrome in 10%.
- In 2007, Akgun.H. et al.,<sup>(15)</sup> compared the frequency of major and minor fetal anomalies reported by second trimester prenatal ultrasound with those found by fetal autopsy after the pregnancy was terminated in the second trimester. Out of 107 cases with major fetal anomalies, 49% showed central nervous system anomalies, 23% had renal and urinary tract anomalies and 11% showed congenital heart diseases. Fetal autopsy was able to confirm all the major anomalies leading to termination of pregnancy that is a 100% success rate. In comparison success rate for prenatal ultrasound was calculated to be 77%. The percentage of extra minor anomalies detected through fetal autopsy was 20%. Also 3% of minor defects indicated by prenatal ultrasonography could not be confirmed by autopsy results.

- Dickinson JE et al.,<sup>(16)</sup> in 2007 shared a 10 year study of perinatal autopsy reviewing the frequency of autopsy following termination of pregnancy for congenital anomalies and its participation in following counseling. For 1012 consecutive terminations for fetal anomalies the major indication were noted to be : abnormal karyotyping in 38.4% cases, neural tube defects in 16.1% fetus, cardiac in 10.3 % cases and cerebral anomalies in 7.5% cases. They performed autopsy in 809 that is 79.9 % of cases. Out of these in 63.5 % cases prenatal diagnosis was confirmed by autopsy with no additional information, lastly, for 15.1% cases significant new information was provided by perinatal autopsy.
- Vogt.C et al.,<sup>(17)</sup> in 2012 carried out a retrospective study on 455 autopsies of fetuses and infants having developmental anomalies after attaining their prenatal ultrasound. The study showed a complete consensus between prenatal ultrasound and postmortem findings in 84% cases, out of these, postmortem examination were supplementary to prenatal ultrasound diagnoses in 16% and in four cases it further helped counseling of the parents. The agreement regarding the main diagnosis made by ultrasound and that of autopsy was made out to be 98% in comparison to 75 % over the previous ten years. The most frequent defects involved in this study included central nervous system, cardiovascular system and urinary tract, respectively.
- In a study done by Godbole K et al.,<sup>(5)</sup> in the year 2013, 141 second-trimester fetuses were studied and a total of 301 structural abnormalities were identified. In 40.4% (57) of the cases, a specific cause was recognised or a syndromic diagnosis was established. The maximum number of systemic anomalies (45/301) belonged to central nervous system. These abnormalities (CNS) were most typically accompanied with facial dysmorphism, including cleft lip and palate. There was a complete concordance between ultrasound and

autopsy findings in 29.07%(41) cases, additional information obtained by autopsy that influenced the final diagnosis was seen in 65 (46.09%) cases.

- In 2014 Nayak S.S. et al.,<sup>(18)</sup> performed the autopsy on 230 fetuses. 106 cases showed single system and 92 cases showed multisystem involvement. Antenatal findings were validated for 23% of cases and additional findings were observed in 37% of cases. In 23% of cases, autopsy findings unearthed new findings and led to change in the final diagnosis. Although, 17% of cases show no cause of fetal loss, in 30.3% of the fetuses the risk of recurrence was apparent and it stayed the same post autopsy; but in 4.8% of cases the diagnosis was modified after carefully considering autopsy findings. Therefore in 36% of cases autopsy findings led to enhancement and changes in recurrence risk. In 77% of cases, autopsy helped couples with prenatal counselling by either confirming antenatal findings (35%), or providing new information/ruling out a diagnosis (42%).
- In 2016 Man.J et al.,<sup>(19)</sup> performed a study on 1064 intrauterine deaths, including early, late intrauterine fetal deaths and stillbirths. Out of these in 40% cases definite cause of death could be identified and 60% were classified as unidentifiable cause. They also discussed that black and asian women had a higher probability of deaths due to ascending infections.
- Cherian.A et al.,<sup>(4)</sup> in 2016 conducted a study aimed to identify the prevalence and types of congenital abnormalities at birth on 36074 births over a period of ten years. It was recorded that 511 were stillbirths and 309 were early neonatal deaths, thus yielding a perinatal death rate of 23 per 1000. Out of total live births, 449 cases were diagnosed with congenital anomalies leading upto a birth incidence of 12.53 per 1000 births. They noted that the commonest anomalies belonged to musculoskeletal deformities (affecting one or a

combination of bone and muscle development in context to skull, trunk or limbs), this was followed by craniovertebral anomalies. The commonest musculoskeletal anomaly comprising of 73.6% cases belonged to congenital talipes equinovarus and among craniovertebral anomalies, meningocele and/ or encephalocele accounting for 36.9% cases, followed by 34% cases of anencephaly.

- Bhide.P et al.,<sup>(20)</sup> in 2016 studied the prevalence of congenital anomalies in an Indian cohort on 2107 pregnant women with outcomes of miscarriage, termination of pregnancy, live or stillbirth. Among 1822 births, the prevalence of congenital anomalies was calculated to be 230.51 per 10000 births. Most common congenital anomalies in the study with prevalence of 65.86 per 10000 births was made to be congenital heart defects. Neural tube defects showed a significant prevalence of 27.44 per 10000 births. They also shared that in their study that congenital abnormalities were the second leading cause for neonatal fatalities. Prenatal diagnosis for congenital anomalies were made in 10.98 per 1000 births and rate for congenital termination of pregnancy was 4.39 per 1000 births.
- In the year 2016 Rossi C et al.,<sup>(6)</sup> conducted the fetal autopsy on 3534 fetuses. They confirmed prenatal ultrasound in 68% (2401) cases, provided additional information in 22.5% (794) cases, and could not confirm prenatal ultrasound in 9.2%(329) fetuses. Additional findings led to change in the final diagnosis in 3.8% of cases. CNS malformations (36.6%) and cardiovascular defects (15.2%) were the most prevalent causes for pregnancy termination or stillbirth. CNS anomalies showed the maximum concordance between ultrasound and autopsy findings (79.4%) followed by termination of pregnancy due to abnormal karyotyping(79.2%), Genitourinary defects(79.9%), skeletal system anomalies (76.6%), Congenital Heart Disease(75.5%), Respiratory system(69.7%),

Gastrointestinal system(62.6%), multiple system (involvement of more than one system)(37%) and limbs(23.3%).

- Struksnaes. C et al.,<sup>(21)</sup> in 2016 studied 1029 cases of termination of pregnancy with gestational age ranging between 11 to 33 weeks. There was a full agreement between ultrasound and autopsy findings in 88.1% of cases with no additional findings and about 97.9% final diagnosis made by ultrasound being correct. In 1.3 % of the cases, autopsy was unable to confirm the ultrasound findings. Also there was no false positive diagnoses that may would have lead to termination of pregnancy.
- In a study done by Venkataswamy C et al.,<sup>(22)</sup> in the year 2018 on 66 fetuses including 17 intrauterine fetal death, and 49 terminations for congenital malformations. The most prevalent anomalies were the central nervous system (neural tube defect), followed by genitourinary system. Except for three cases, autopsy findings was able to confirm prenatal ultrasound findings in all cases. A full consensus between ultrasound and autopsy was established in 39.7% (17) cases. In 62.2% (25) cases additional findings were noted, among these, 15 cases had a significant change of recurrence risk due to alterations in initial ultrasound diagnosis.



## MATERIALS AND METHODS

### **1. Study Design**

**1.1 Study setting:** The study was conducted in the Histopathology section, Department of Pathology, BLDE (Deemed to be University), Shri B.M Patil Medical College, Hospital, and research center, Vijayapura.

**1.2 Study type:** A hospital-based cross-sectional study design to evaluate fetuses sent to histopathology section for the purpose of fetal autopsy after spontaneous abortions or medically terminated in our hospital and other hospitals referred to our department.

**1.3 Study period:** The study was conducted from 1<sup>st</sup> April 2023 to 31<sup>st</sup> December 2024.

**1.4 Ethical clearance:** Ethical clearance for this study was received from Institutional Ethical Committee/ BLDE (Deemed to be University) BLDE (DU)/IEC/939/2023-24.

**1.5 Sample size:** Sample size was calculated on the basis of anticipated proportion of complete agreement between ultrasound and autopsy findings of 29.07%, the study required a sample size of 80 fetuses with a 95% level of confidence and 10% absolute precision.

Formula used

- $$n = \frac{z^2 \cdot p \cdot q}{d^2}$$

Where Z= Z statistic at  $\alpha$  level of significance

$d^2$ = Absolute error

**P= Proportion rate**

$q= 100-p$

- To increase the efficiency of present study the final sample size collected was 150 fetuses.

## **1.6 Statistical analysis**

- The data obtained was entered into a Microsoft Excel sheet, and statistical analysis was performed using a statistical package for the social sciences ( Version 20).
- Results were presented as percentages, and diagrams.
- JMPSAS SOFTWARE was used for this statistical analysis.

## **2. Sample Recruitment**

**2.1 Inclusion criteria:** All fetuses sent to the histopathology section with the objective of autopsy.

**2.2 Exclusion criteria:** Autolysed fetuses and fetuses without ultrasonographic reports.

**2.3 Consent and confidentiality:** Before inclusion in the study, all participants were informed regarding the objectives, procedures, potential benefits and risks of the study. Written informed consent was obtained from each participant in the study.

## **3. Sample Collection**

**3.1 Protocol for fetal examination:**

The fetal pathology examination follows a similar pattern of evaluation as that of adults, but differ in certain important aspects, such as, head examination which includes special problems like interpretation of developmental changes, recognition of malformations and evaluation of injuries during birth. <sup>(8,23)</sup>

The fetus was received and fixed in 10% formalin. 100% that is concentrated formalin was injected through anterior fontanelle in the fetus brain. The duration of brain fixation was 7 to 10 days. <sup>(23,24)</sup>

### **3.2 Radiographic examination:**

Whole body antero- posterior and lateral radiographs were taken whenever indicated especially when bony abnormalities were suspected.

### **3.3 External examination:**

The external examination included weighing and taking measurements of the fetus, including : head circumference (HC), chest circumference at nipple level (CC), crown- rump length (CRL), abdominal circumference at umbilical level (AC), crown- heel length (CHL), inner intercanthal distance foot length. Foot length was taken in every case and used to confirm the gestational age as mentioned on ultrasound.

The study consists of records with reference to clinical history which included details of maternal health, family history, past and present obstetric history, antenatal care and ultrasonographic investigations.

**Table 2: Foot length chart:** <sup>(25)</sup>

WEEKS	FOOT LENGTH (mm)
12	14
14	20
16	27
18	33
20	39
22	45
24	50

### **3.4 Photographs:**

Photographs were taken for every case. The photographs taken depicted the abnormal features. In situ photographs were also taken, were ever indicated for preserving anatomic relationships and depict initial presentation of visceral lesions <sup>(23)</sup>.

### **3.5 Equipments:**

Special instruments suitable for the purpose of fetal autopsy, including pediatric surgery tools were used.

### **4. Initial incision:**

A Y- shaped incision was made, the arms of the Y incision was extended to the top of the shoulders for the purpose of freeing up the skin just above the anterior aspect of the neck and inferiorly in the midline, at the level of xiphoid process. The attachment of skin flap to the chest wall was incised and pulled upwards over the chest. Initially a midline vertical incision was made extending from the xiphoid process up to the symphysis pubis by incising around the left side of umbilicus. The abdominal organs were first inspected. The attachments of mesentery along with the location of the appendix were examined <sup>(9, 23)</sup>.

### **5. In -situ examination:**

#### **5.1 Thorax:**

An upside-down V shaped incision was made with the objective to remove chest plate. The above incision began at sternoclavicular joint, 4mm from the costochondral junction. The xiphoid process was grasped with forceps and the ribs were removed away from the thoracic organs, following this fibrous attachments were incised very close to the bones. and fibrous attachments were cut as close to bone as possible. Lungs were also examined <sup>(9, 23)</sup>.

To expose the great vessels and heart the pericardium and thymus were removed together. After nicking the pericardium, a cut parallel to the diaphragm was made that further extended till the pulmonary veins (PV) on the left side and the base of inferior vena cava (IVC) on the right side, respectively. Then the pericardium was incised on the right in close proximity to IVC, up to the level of left innominate vein following the lateral side of right atrium (RA) and superior vena cava

(SVC). Meanwhile, on the other side, the scissors were positioned at ninety degree to the diaphragm at the level of the PVs as they leave the left atrium (LA). For proper visualization, a continued cut was made adjacent to the left pulmonary artery. The thymus was then dissected away from pericardium<sup>(9, 23, 24)</sup>.

## **5.2 Heart:**

A thorough and comprehensive evaluation of the exterior appearance of the heart and all vascular connections were made to rule out any congenital anomalies. The location of the heart and the direction of its apex in the chest was noted following this inspection of the great arteries was performed. The position of the pulmonary trunk with respect to aortic trunk was also noted<sup>(9, 26)</sup>.

## **5.3 In -situ opening of heart:**

In -situ opening of heart was carried out by using coronary arteries as a guide and following the flow of blood, avoiding the septum. The following basic steps were followed:

1. The initial nick was made lateral to the RA. Scissors were inserted to open the SVC and this cut was continued to left innominate vein. Following this the IVC was opened inferiorly up to the diaphragm<sup>(26)</sup>.
2. Using forceps the RA wall was lifted and one end of the scissors was introduced to the RA and the inferior aspect of RA wall was cut. Tricuspid valve along with RA were then meticulously examined. Using posterior descending artery (PDCA) as a guide, the cut was then extended across the tricuspid valve (TV) to the RV apex. The RV was then inspected<sup>(26)</sup>.
3. An incision was made using anterior coronary descending artery (ACDA) as guide for RV starting from the apex across pulmonary valve and continuing up to left pulmonary artery<sup>(26)</sup>.
4. LA appendages were nicked at the tip and scissors were inserted, extending the cut into each of the pulmonary veins. Mitral valve examined<sup>(26)</sup>.

5. Using the PDCA as a guide, the LV was incised postero- laterally from the MV till the apex.

From the apical portion this cut was extended from the anterior wall of the LV to the aortic valve, using the ADCA as a guide. To separate the pulmonary trunk from the ascending aorta we performed a blunt dissection. For opening aortic valve, ascending aorta and the aortic arch the final blunt dissection cut was made <sup>(26)</sup>.

## **6. Removal of organs:**

### **6.1 Spinal cord: Anterior approach:**

Following evisceration of all organs there is a full view of thoracic and lumbar portions of spinal cord. Lower most lumbar intervertebral discs were then transected using a scalpel and one end of the rounded pair of scissors was inserted into this opening to make a continuous cut. A cut was made between the dura and the bone, being careful that the dura was left intact. After dissecting all the spinal pedicles, vertebral bodies were lifted, exposing the spinal cord. Using a sharp scalpel blade, the cord was transected at the lumbar end and with the help of toothed forceps the surrounding dura was gently lifted. Without exerting any tension to the cord the dura and spinal cord was dissected from the lumbar to cervical portion <sup>(27)</sup>.

### **6.2 Brain:**

The incision on the skin was extended from behind one ear, upwards, over the top of the cranium and then down behind the other ear. The skin flap was reflected anteriorly over the eyes and posteriorly in the caudal direction. The length and breadth of fontanel were measured. Following which on each lateral corner of the anterior fontanel a small nick was made with the purpose of opening the skull. A complete oval cut was made by inserting rounded scissors into the nick of each side, leaving a portion unmarred on the lateral side that acts as a hinge allowing the bony flap

to be folded away from the brain. Then lateral to the sagittal sinus incisions were made through the bone to leave it intact. The brain was then inspected in -situ. Using a sharp scalpel the bony attachments was made free from the falx and the tentorium<sup>(9, 27)</sup>.

For removing the brain, our left hand was positioned on top of the occiput, then to ensure that the brain is not detached from the skull bone the brain and skull was cradled in the palm. The head was gently tilted back to allow the brain to fall away and separate from the calvarium on its own. The index and the middle finger of the right hand were used to gently retract the cerebral hemisphere. Following this the optic nerves were cut close to the skull. The bottom surface of the anterior spinal cord and brainstem were now evidently visible. The cervical spinal cord was transected and separated as far as possible<sup>(9, 27)</sup>.

### **6.3 Intact Brain and Spinal cord: Posterior approach:**

For preservation of skull and brain anomalies this approach was used, including anomalies such as occipital encephalocele, Arnold-chiari malformation, Dandy-walker syndrome and meningocele anywhere along the spinal length. The incision in the skin was in the form of a question mark (?). The portion that is extended on top of the neck was further extended caudally as far as the defect needed to be preserved. The skin over the skull was reflected. The muscle over the occiput was carefully removed and the soft tissues over the rami of upper cervical vertebrae were dissected away. If needed the atlas was cut along the second and third cervical vertebrae. A careful incision was made along the dura for the purpose of exposing cervical cord and foramen magnum. Under normal conditions the cavity of fourth ventricle and the cerebellar tonsils were seen. A pair of blunt scissors were placed between the dura and the bone and a cut was made on either side to allow removing the spinal cord, with or without a spinal defect. A cut was also made for removal of bone around the defect. An additional cut in the midline of the occipital plate was



made and the brain was removed as described earlier this allowing the brain and cord to be removed together<sup>(9, 27)</sup>.

## **7. Evisceration:**

Rokitansky technique was followed for evisceration, which allowed removal of organs of the neck, chest and abdomen as a single unit. Organs were then separated from each other and the weight of individual organ was taken<sup>(9, 24)</sup>.

### **• Organ block Dissection:**

The organ block was positioned on the ventral surface.

The ventral surface of the organ block was placed facing downwards. The aorta was opened posteriorly to the aortic arch. Then renal arteries were opened, leaving them attached to the kidneys and for separating either side of the kidney a segment of aorta was cut. The diaphragm was reflected away from the adrenals and each adrenal was removed alongside either side of kidney in their anatomical position. To preserve the ureters, the kidneys were dissected away from the block. For female fetuses urinary bladder and external genitalia were dissected free from the rectum, unless indicated otherwise in case of an anomaly. The kidneys and adrenals of respective literalities were weighed together<sup>(9, 23)</sup>.

The bile duct was opened, and gall bladder was gently squeezed to allow some bile to pass through for confirmation of patency of cystic duct and examine porta hepatis. Then the portal vein and hepatic artery were opened. The splenic vein was also opened and the spleen was removed. Perisplenic fat was examined for presence of accessory spleen and weight of spleen was recorded. The intestines were cut at the duodenum after carefully getting separated from mesentery<sup>(9, 23, 24)</sup>. The intestine was then opened, the contents were emptied and mucosa was thoroughly examined. The esophagus was reflected from the trachea after opening it, to eliminate the chances of

transecting a fistula. After dissecting the trachea away from the esophagus, the diaphragm was nicked away to remove a single block containing the esophagus, stomach, duodenum and pancreas. Then from the block, opening of stomach and duodenum was carried out, by cutting the stomach along the greater curvature and sectioning of pancreas was done. Then the liver and diaphragm were separated from the thoracic organs and the liver and diaphragm were dissected free from each other. The liver was weighed and the gall bladder was opened <sup>(9, 24)</sup>.

The liver and lungs were dissected from each other and separately weighed. Subsequently, the trachea was opened into each big bronchus and posteriorly to the carina to carefully assess the bronchial morphology. Thereafter, lungs were sectioned and following the flow of blood the heart was examined again. At this moment, congenital anomalies of the heart were demonstrated by the process of windowing that is by carefully removing portions of myocardium or vascular connections for the purpose of photography <sup>(9, 24)</sup>.

The brain was examined after 7 to 10 days of fixation. External surface was examined for any abnormality like malformations, hemorrhage that may be overlooked in fresh state <sup>(27)</sup>.

### **Sectioning the fixed brain:**

The fixed brain was washed in running water overnight. A thorough examination of the dura, falx and the tentorium was conducted and the dural sinuses were opened. The leptomeninges were examined. The general appearance and symmetry of the cerebral hemispheres were noted. The degree of development of cerebral convolutions were recorded along with the general appearance and size of the cerebellum. We also identified the components of the brain stem. The cerebrum was separated from the cerebellum and brainstem by cutting through cerebral peduncles as far rostrally as possible. The cerebrum was placed upside down (base up) on the cutting board. It was sliced in serial coronal sections beginning at the frontal pole and ending at the occipital pole using a brain

knife, the first cut was made at the level of the mammary bodies. The thickness of the slices was determined by the consistency of the brain. Firm brains were sliced at 1 cm intervals, soft friable ones at larger intervals. The size and shape of the ventricles were noted <sup>(9, 27)</sup>.

Using a scalpel a cut through cerebellar peduncle was made and the brain stem was separated. The cerebellum was sectioned serially beginning at the superior surface in the horizontal plane using a knife. Anatomic landmarks were also identified. The brainstem was sectioned serially from rostral to caudal ends at 2 mm intervals <sup>(9, 27)</sup>. Finally, the size of fourth ventricle and aqueduct of sylvius were noted <sup>(27)</sup>.

## **8. Microscopic Examination:**

Gross findings were sufficient for diagnosis of a majority of cases, yet microscopic examination was conducted with hematoxylin and eosin slides <sup>(9)</sup>.

The sections submitted for microscopic examination of various organs were as follows: lungs (one section from each lobe), heart (two sections), liver (one section from right and left lobe respectively), one section from the spleen, adrenals (right and left each), kidneys (right and left each), thymus and umbilical cord. In few special cases where indicated, sections from pancreas, urinary bladder, testis or ovary, uterus, vagina, costochondral junction and from any other organ or tissue that appeared abnormal were submitted. Two sections from the placenta where ever indicated were also taken <sup>(8)</sup>.

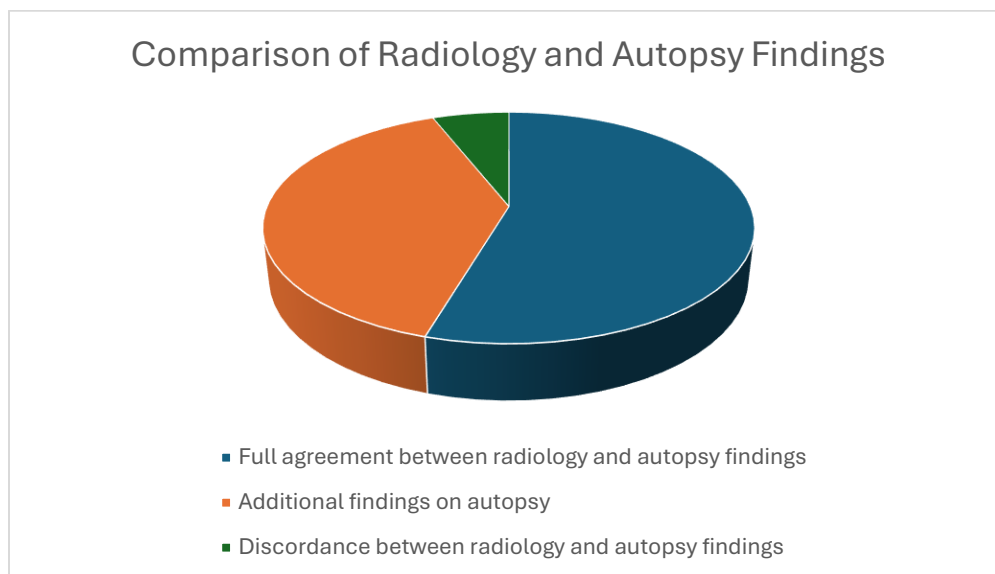
### **OBERVATIONS AND RESULTS:**

The data included observations made in 150 fetuses which are as follows:

**Table 3: Comparison of ultrasound and autopsy findings:**

Sr.No.	Cases	No. of cases	Percentage
1.	Full agreement between radiology and autopsy findings	82	54.6%
2.	Confirmation and additional findings with autopsy	59	39.4%
3.	Discordance between radiology and autopsy findings	09	6%

**Figure 2: Graphical representation of comparison between USG and autopsy findings**



As seen in table 3 and figure 2 there was a complete concordance between ultrasound and autopsy findings in 54.6% (82 out of 150) cases. The autopsy provided additional findings in 39.4% (59 out of 150) cases.

In 68% (40 out of 59) cases additional findings were discovered but it did not lead to classification of fetus in any syndrome or association including low set ears, single umbilical artery, occipital bulge, webbed neck etc.

In our study a total of 23 syndromes (figure 3) were observed. A complete concordance of ultrasound and autopsy were seen in 04 of these suspected syndromic cases with no additional findings.

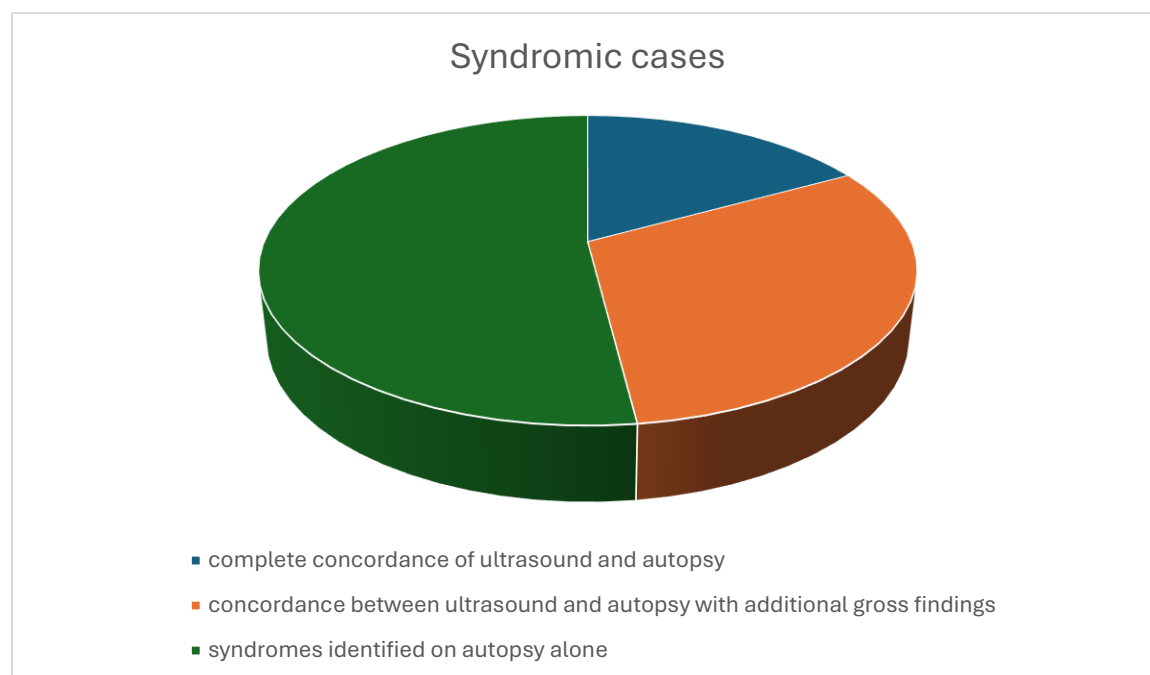
In 20% (12 out of 59) cases no syndromic anomaly was detected on ultrasound and diagnosis was made solely on the basis of additional findings discovered on autopsy classifying these fetuses into a syndrome or association. These findings included cystic medulla and hypoplastic muscle fibres in case one, short neck, bell shaped thorax, hemivertebrae, pleural effusion and depressed nasal bridge in case two, hypoplasia of thymus and low set ears in case three leading to diagnosis of complex congenital anomaly in all three cases. We found additional findings of micromelia, thin elongated ribs and short limbs in case four that led to diagnosis of Greenberg dysplasia. In case five we additionally uncovered bilateral club foot and dextrocardia classifying it as PAGOD syndrome. We discovered presence of spina bifida in case seven, classifying the fetus into anencephaly- spina bifida complex. Case eight was classified as Hydrolethrus syndrome due to additional finding of cleft lip. In case nine additional findings of Right toe polydactyly, bilateral CTEV, scoliosis, reduced right ventricular volume and right ventricular hypertrophy, X- ray showing butterfly anomaly, segmental anomaly all lumbar vertebrae with widening and splaying of anterior elements suggestive of lumbar scoliosis (figure 16), leading to diagnosis of VACTERL

association (figure 14 and 15). An additional finding of dilated ventricles thinned out cortex, absent vermis and bilateral low set ears leading to a diagnosis of Joubert syndrome in case ten. In case eleven we suspected presence of pulmonary alveolar proteinosis which is a rare lung disorder, this diagnosis was confirmed on microscopy by presence of alveolar spaces filled with granular, eosinophilic periodic acid-schiff positive, lipoprotein material. In case twelve we unearthed decreased intra orbital distance, hypoplastic face and prefrontal edema that led to diagnosis of Larsen syndrome (figure 12).

In 12% (07 out of 59) cases a syndrome was suspected in ultrasound and autopsy confirmed the diagnosis along with discovering new gross features but not changing the final diagnosis. The findings included coiled umbilical artery, single umbilical artery, short neck etc.

It should also be noted that ultrasound findings of 02 cases showed presence of a syndrome that is Dandy walker syndrome which could not confirmed.

**Figure 3: Graphical representation of Syndromic cases**



**Table 4. Main diagnosis classified according to organ system involvement (n=150)**

Sr. No.	System involved in anomalies	No. of cases	Percentage
1.	Central Nervous System	34	22.6%
2.	Skeletal system	16	10.6%
3.	Cardiovascular system	13	8.6%
4.	Respiratory system	05	3.3%
5.	Renal system	04	2.6%
6.	Gastrointestinal system	04	2.6%
7.	Lymphatic system	03	2%
8.	Anal canal	03	2%

**Figure 4: Graphical representation of most common systems involved**

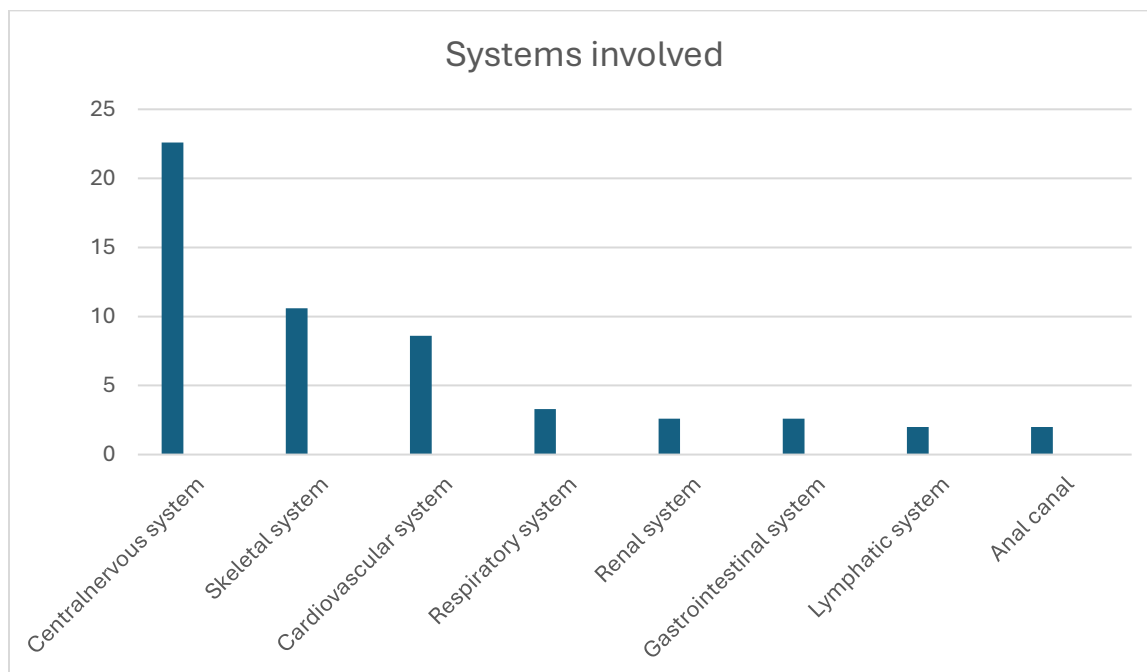


Table 4 and figure 4 signifies that malformations of central nervous system were commonest accounting for 22.6% (34/ 150) cases followed by skeletal system and cardiovascular system, respectively.

**Table No.5 Classification of Central nervous system anomalies. (n=34)**

Sr. No.	Anomaly	No. of cases	Percentage
1.	Meningomyelocele	06	17.6%
2.	Ventriculomegaly	05	14.7%
3.	Hydrocephalus	05	14.7%
4.	Anencephaly	05	14.7%
5.	Arnold chiari malformation	04	11.7%
6.	Hemivertebrae	03	8.8%
7.	Dandy walker syndrome	02	5.8%
8.	Anencephaly- spina bifida complex	02	5.8%
9.	Others	05	14.7%
	Encephalocele	01	
	Corpus callosum agenesis	01	
	Cystic medulla	01	
	Spina bifida	01	
	Moulding of skull bone	01	



**Figure 5: Graphical representation of central nervous system anomalies**

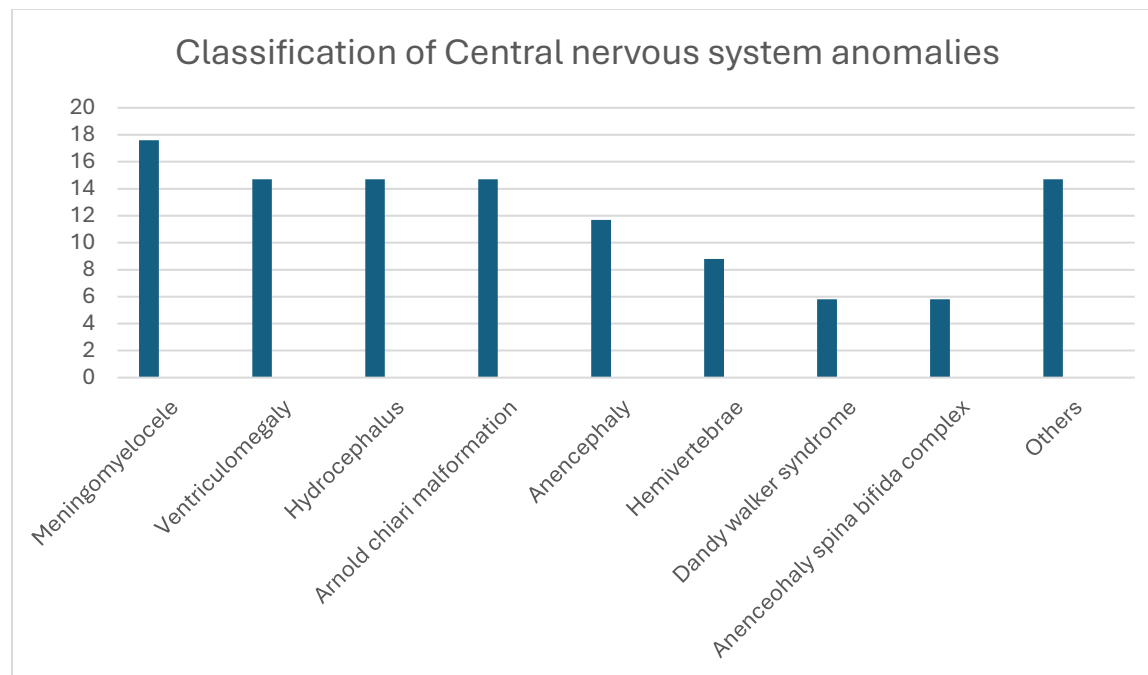
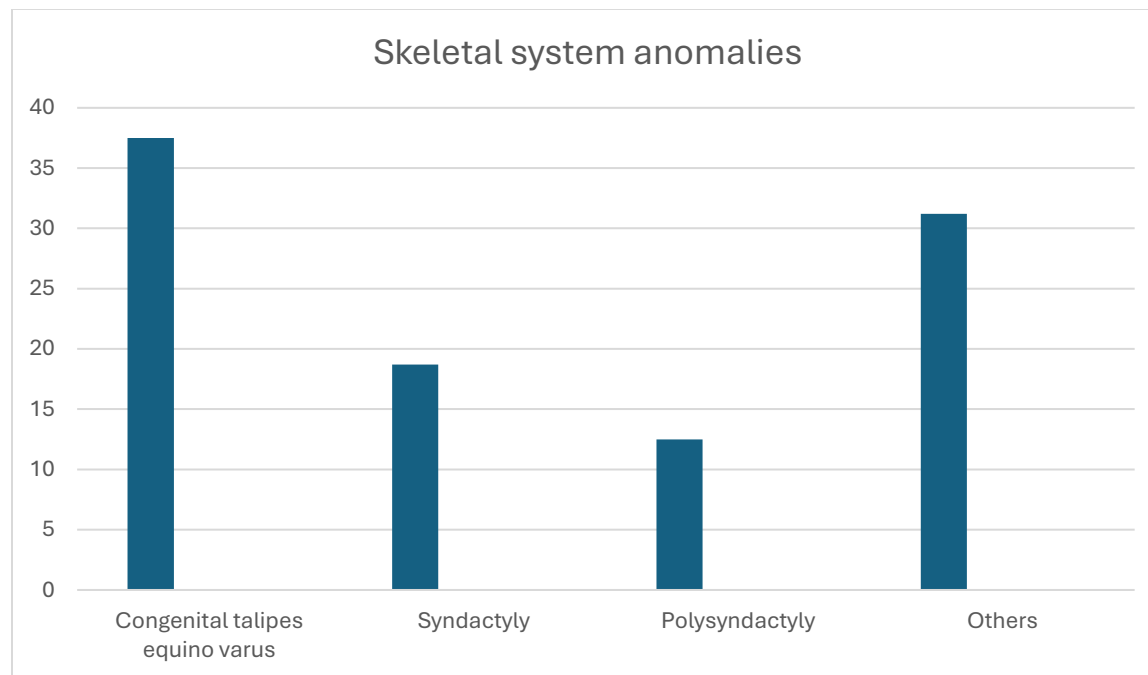


Table 5 and figure 5 shows that the commonest anomaly in relation to central nervous system is neural tube defect which includes 6 cases of meningomyelocele (figure 21), 4 cases of anencephaly (figure 22), 1 case of encephalocele, 2 cases of anencephaly- spina bifida complex and 1 case of spina bifida. The second most common malformation was that of ventriculomegaly, hydrocephalus and Arnold Chiari malformation all three seen in 5 patients, respectively. It is also noted worthy that two cases of Dandy walker syndrome were encountered. It is also worth mentioning that 01 out of the 05 diagnosed cases of hydrocephalus was accompanied by polysyndactyly and cleft lip, both features observed on gross which lead to the diagnosis of Hydroletharus syndrome solely on autopsy findings.

**Table No. 6: Classification of skeletal system anomaly (n=16)**

Sr. No.	Anomaly	No. of cases	Percentage
1.	Congenital Talipes Equino Varus/ Valgus	06	37.5%
2.	Syndactyly	03	18.7%
3.	Polysyndactyly	02	12.5
4.	others	05	31.2%
	Left foot talipes	01	
	Decreased stature	01	
	Short limbs	01	
	Contractures in upper and lower limb	01	
	Thin elongated bones	01	

**Figure 6: Graphical representation of skeletal anomalies**



As shown in table 6 and figure 6 there were a total of 16 cases with skeletal anomalies, with maximum cases of congenital talipes equino valgus/ varus. It should also be noted that a fetal X-ray was done in most cases to confirm and support our findings. One of the cases of congenital talipes equino valgus/varus came along side other findings of dilated ventricles, thinned out cortex, absence of vermis and low set ears pointing towards hydrocephalus thus coming to a diagnosis of Joubert syndrome which is caused by faulty genes that prevent the cerebellar vermis from fully developing.

**Table No. 7: Classification of Cardiovascular anomalies (n=11)**

Sr. No	Anomaly	No. of cases	Percentage
1.	Hypoplastic left heart syndrome	02	18.1%
2.	Dextrocardia	02	18.1%
3.	Tetralogy of fallot	02	18.1%
4.	Right aortic arch	02	18.1%
5.	Atrioventricular septal defect	02	18.1%
6.	Others	04	36.3%
	Persistent left superior vena cava	01	
	Dysplastic mitral valves		
	Right ventricular hypertrophy	01	
	Pulmonary stenosis	01	
		01	

**Figure 7: Graphical representation of cardiovascular anomalies**

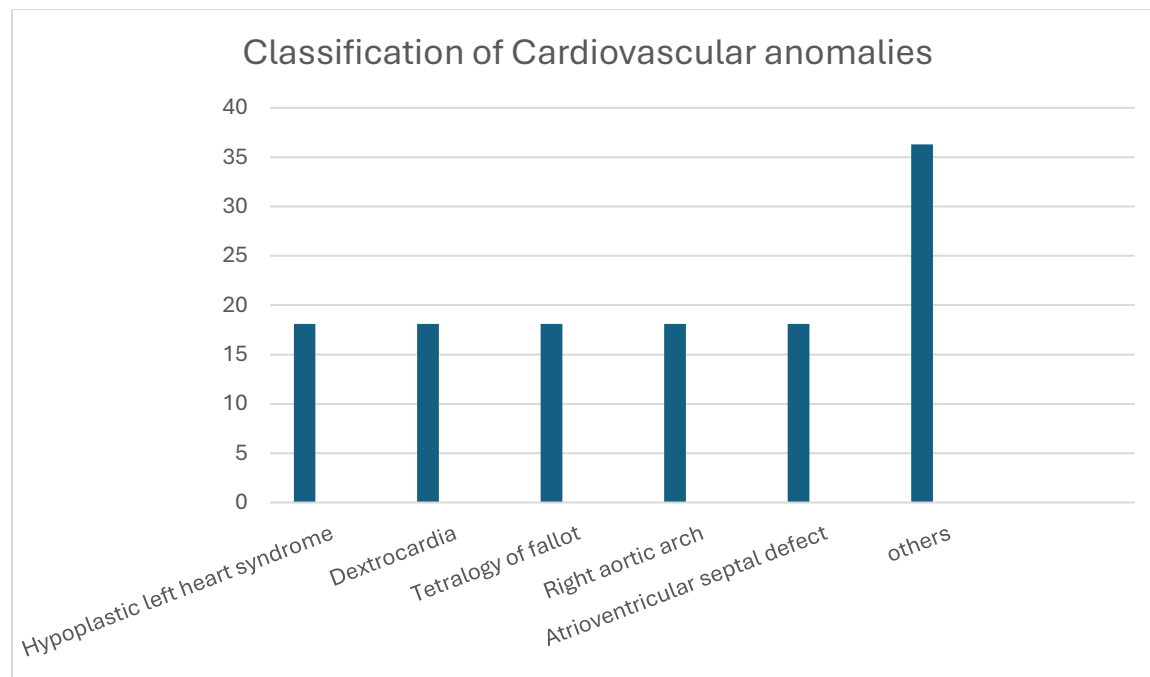


Table 7 and figure 7 shows, hypoplastic left heart syndrome as the commonest heart syndrome defect encountered along with other anomalies like dextrocardia (figure 19), tetralogy of fallot, right aortic arch and atrioventricular septal defect. All of the above mentioned anomalies were observed in 02 out of 13 cases, respectively.

**Table No. 8 Classification of Respiratory system Anomalies (n=5)**

Sr. No	Anomaly	No. of cases	Percentage
1.	Pulmonary alveolar proteinosis	01	20%
2.	Blake pouch cyst	01	20%
3.	Tracheal deviation	01	20%
4.	Left lung hypoplasia	01	20%
5.	Pleural effusion	01	20%

**Figure 8: Graphical representation of respiratory system anomalies**

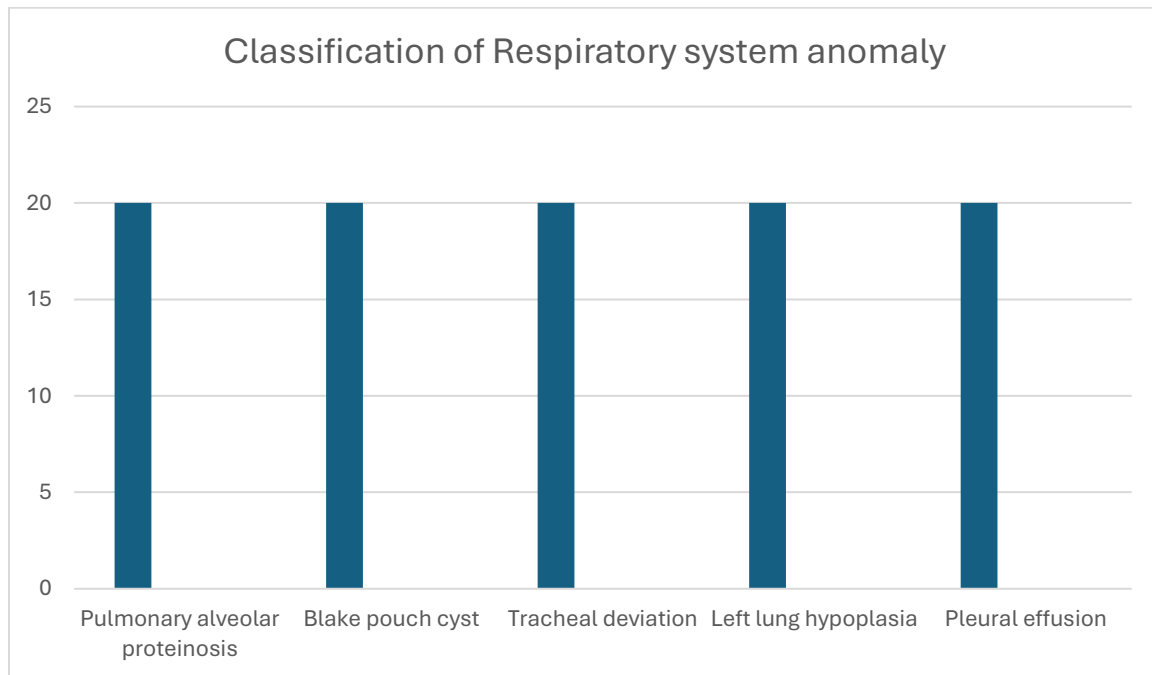


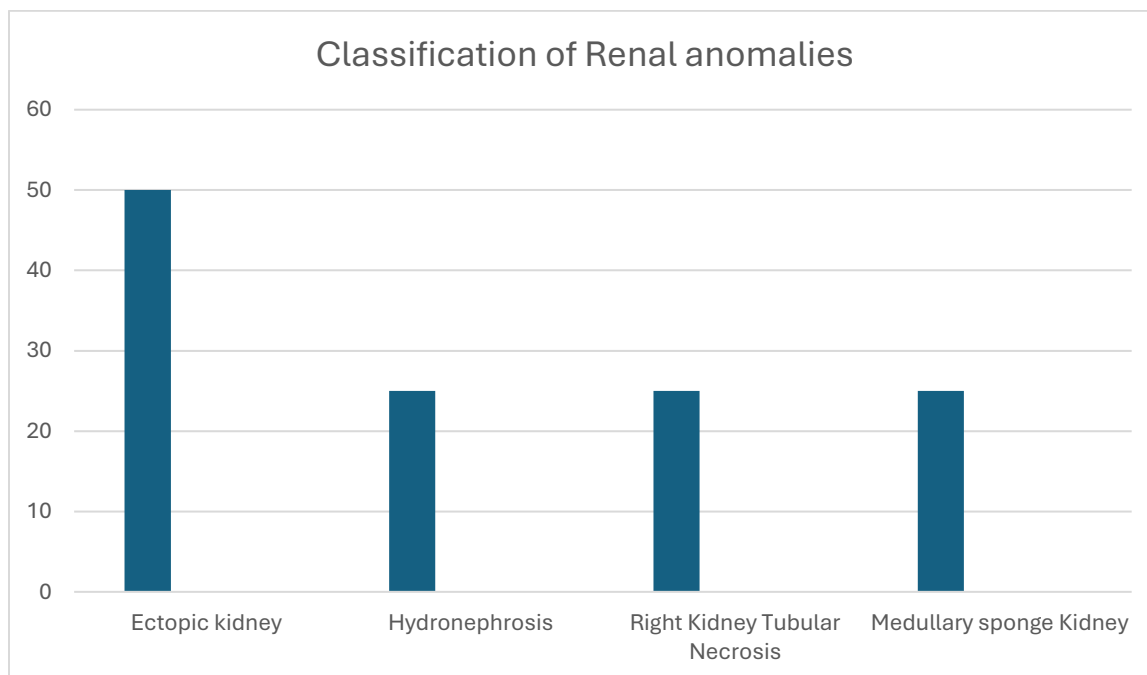
Table 8 and figure 8 represents the common anomalies encountered in respiratory system. The only case of left lung hypoplasia was accompanied by other findings including Dextrocardia,

diaphragmatic hernia and bilateral club on gross which lead to its reclassification in PAGOD syndrome. PAGOD syndrome stands for pulmonary hypoplasia- agonadism- dextrocardia- diaphragmatic syndrome, which is a severe developmental syndrome.

**Table No 9: Classification of Renal anomalies (n=4)**

Sr. No.	Anomaly	No. of cases	Percentage
1.	Ectopic kidney	02	50%
2.	Hydronephrosis	01	25%
3.	Right kidney tubular necrosis	01	25%
4.	Medullary sponge kidney	01	25%

**Figure 9: Graphical representation of renal anomalies**



The table no. 9 and figure 9 shows that ectopic kidney was seen in 02 cases. In one case of ectopic kidney it was accompanied by hydronephrosis. There was a single case of medullary sponge kidney that was associated with Arnold Chiari malformation.

**Table 10: Classification of Gastrointestinal Anomalies (n=4)**

Sr. No.	Anomaly	No. of cases	Percentage
1.	Gastrochisis	02	50%
2.	Inversus siatus	01	25%
3.	Herniation of stomach into hemithorax	01	25%



**Figure 10: Graphical representation of gastrointestinal anomalies**

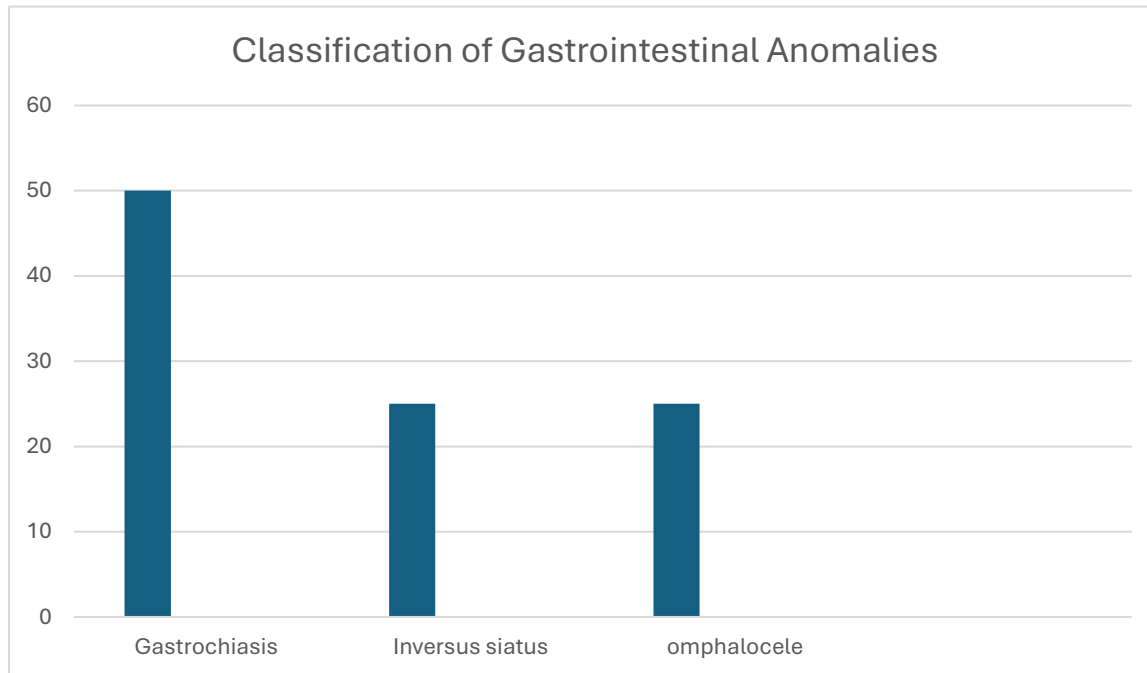


Table 10 and figure 10 shows the distribution of gastrointestinal anomalies cases seen in our study. It is noteworthy that one out of the two cases of gastrochiasis was accompanied by diaphragmatic hernia, dextrocardia and edematous external genitalia that lead to the diagnosis of complex congenital anomaly for that fetus. We also encountered a single case of omphalocele (figure 18).

**Table No. 11: Facial anomalies (n=25)**

Sr. No.	Anomaly	No. of cases	Percentage
1.	Low set ears	15	60%
2.	Cleft lip	03	12%
3.	Flat head	02	8%
4.	Cleft palate	02	8%
5.	Occiput protuberance	02	8%
6.	Others	04	16%
	Flat nasal bridge	01	
	Hypertelorism	01	
	Underdeveloped ears	01	
	Globular head	01	

**Figure 11: Graphical representation of facial anomalies**

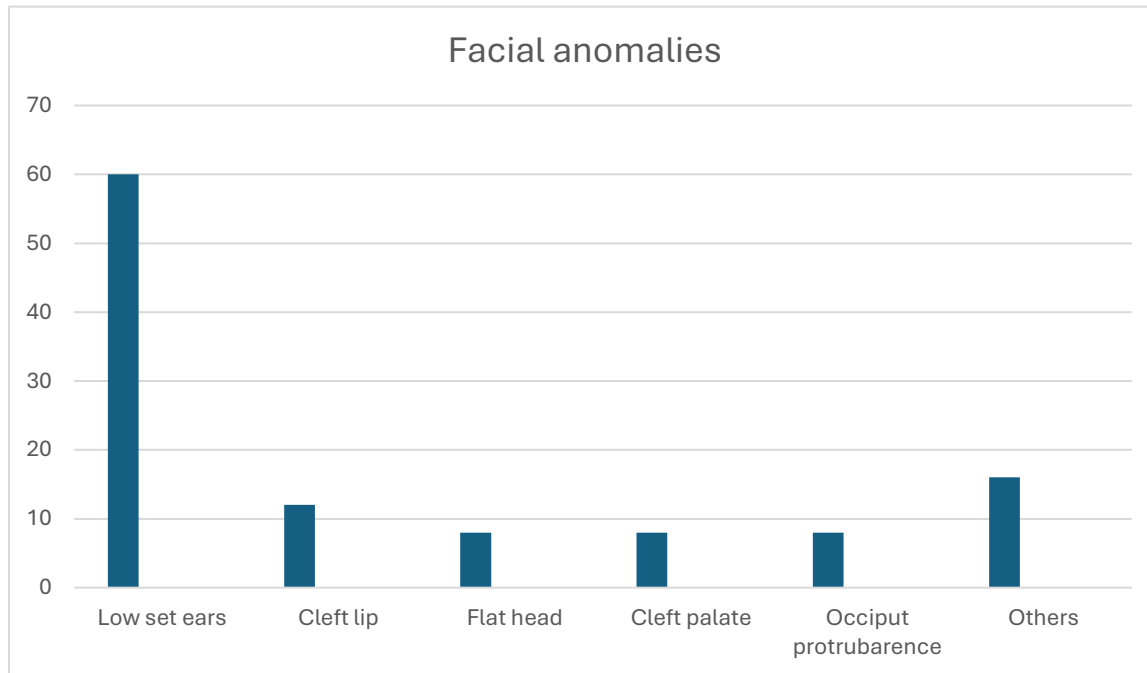


Table 11 and figure 11 shows that among all the facial anomalies seen, low set ears were the most common that is seen in 15 out of 25 cases (60%) followed by cleft lip in 03 out of 25 cases (12%) (figure 25). Low-set ears are defined as ears positioned below the horizontal plane from the outer canthus that is the corner of the orbits straight back to the occiput .

Out of 150 cases 03 cases were showing evidence of cystic hygroma which is a benign birth defect that occurs due to malformation in lymphatics. Cystic hygroma was the only lymphatic malformation seen. Although, one of the cases of cystic hygroma was accompanied by external micromelia that is shortening of both proximal and distal bones equally, short limbs and thin elongated ribs leading to diagnosis of Greenberg dysplasia on gross.

Imperforate anus was detected in 02 out of 150 cases and 02 cases with twin to twin transfusion syndrome showed hypolobated right lung (figure 17).

Ultrasound findings also showed presence of Harlequinn syndrome in 01 case which was confirmed on gross by presence of thick, cracked skin plates and fissures (figure 23) and on histopathology of skin showing hyperkeratosis and abnormal keratotic material around hair shaft (figure 24).

We also came across 03 cases of fetuses with short neck also known as pterygium colli (figure 20) that denotes a congenital condition with additional skin fold extending from neck to shoulders.

## PHOTOMICROGRAPHS



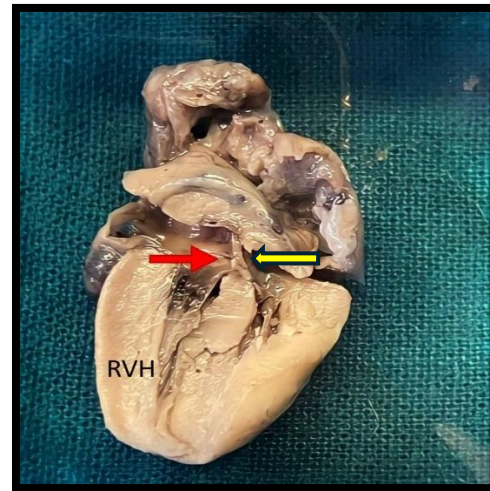
**Figure 12 a) and b) Case of Larsen syndrome:** Gross showing Frontal bossing, flattened mid face, hypotelorism, low set ears, long and spatulate fingers, elbows and hips flexed, hyperextended knees, club foot.



**Figure 13: X- ray findings in Larsen syndrome:** Underdevelopment of fetal calvarium and sacrum, right sided short arm suggestive of congenital dysplasia, hyperextended bilateral knees and rocker bottom feet.



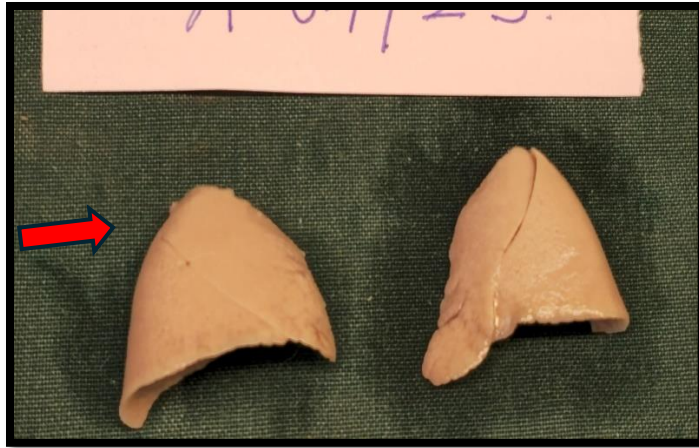
**Figure 14: Case of VACTERL Association:** Right talipes equino varus, left talipes equino valgus and right toe polydactyly (red arrow).



**Figure 15: VACTERL Association** showing right ventricular hypertrophy, overriding of aorta (yellow arrow), incomplete ventricular septum (red arrow), pulmonary trunk not identified.



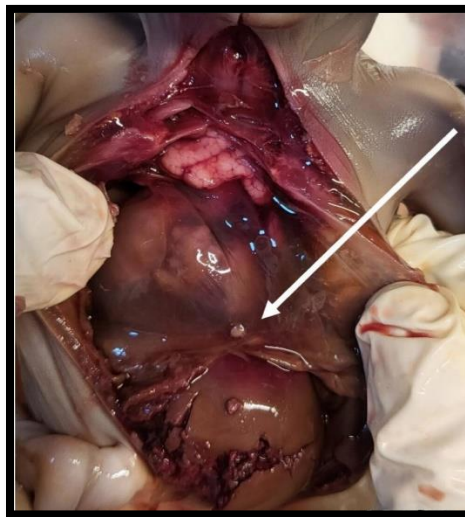
**Figure 16: X- ray findings for VACTERL Association:** D11-D12 butterfly vertebrae with widening and splaying of anterior elements (white arrow).



**Figure 17:**Hypolobated right lung (red arrow)



**Figure 18:**Omphalocele- abdominal contents protruding out of umbilical cord (yellow arrow).



**Figure 19:**Dextrocardia (white arrow)



**Figure 20:** Webbed neck  
anomalous baby (red arrow)



**Figure 21:** Anomalous baby with spinal  
defect suggestive of meningocele  
(red arrow).



**Figure 22:** Anomalous baby showing  
anencephaly (red arrow) and herniation  
of bowel loops from abdomen (yellow  
arrow).





**Figure 23:** Harlequinn baby syndrome



**Figure 24:** Harlequinn syndrome baby skin histopathology showing hyperkeratosis and abnormal keratotic material around hair shaft.



**Figure 25:** Anomalous baby with cleft lip (red arrow) and palate.

## **DISCUSSION**

- Fetal loss has a profound and complex psychosocial impact on parents and leads to grief and anxiety. Following this loss fetal autopsy examination plays a major role not only to confirm the prenatal findings and provide a sense of relief to the parents but also to help counsel them on the likelihood of recurrence for future pregnancies <sup>(28)</sup>.
- Our study included fetuses that were either medically terminated due to structural anomalies detected in prenatal scans or underwent spontaneous abortion. On carefully comparing the autopsy findings and ultrasound of the fetuses, there was complete concordance in 82 cases (54.6%). In 59 cases (39.4%) autopsy led to unmasking of additional findings that were not mentioned in ultrasound. In the remaining 09 cases (6%) there was lack of agreement between the ultrasound findings and autopsy findings, which could be attributed to autolysed brain, inability to analyse petite anomalies that were difficult to visualize with naked eye or due to error in interpretation of ultrasound report. These findings were comparable to studies done by Isaken C.V et al.,<sup>(29)</sup> and Rossi A.C et al.,<sup>(6)</sup>
- The 59 cases with additional findings in our study included 40 cases where these findings did not impact the final diagnosis made on ultrasound. In 07 cases ultrasound findings had a suspicion of presence of a syndrome and autopsy confirmed the same. In 12 out of these 59 cases, these additional findings helped in identifying syndromes that were suspected on ultrasound, thereby making their diagnosis possible solely based on autopsy findings.
- In the present study, a total of 23 cases (15.33%) were classified into a syndrome, these included 04 cases where the ultrasound findings and autopsy findings were in complete concordance, that is the suspected syndrome on ultrasound was confirmed by autopsy with no additional findings. In 07 cases autopsy inspection led to uncovering of few additional findings

without changing the final diagnosis of syndrome made by ultrasound. Out of these 23, 12 cases did not show presence of any syndrome in ultrasound, but the intensive and articulated autopsy of these cases lead to additional findings finally leading to their classification into a syndrome. It is noteworthy that ultrasound of 13 fetuses showed presence of a syndrome, but autopsy could not confirm this finding. This presence of disagreement between ultrasound and autopsy is majorly attributed to the fact that both the cases showed presence of syndrome involving central nervous system and in the both the cases the brain was autolysed that acted as an obstacle in confirmation of the ultrasound findings.

- In the present study the most common system involved was central nervous system consisting of 34 (22.6%) out of 150 total cases. This was followed by involvement of skeletal system (10.6%), cardiovascular system (8.6%), respiratory system (3.3%), renal and gastrointestinal system (2.6%) and lymphatic system (2%). We also encountered presence of imperforate anus in 03 out of 150 cases (2%). These results were comparable to studies done by Vogt. C et al.,<sup>(17)</sup> Struksnaes. C et al.,<sup>(21)</sup> Godbole. K et al.,<sup>(5)</sup> Nayak S.S et al.,<sup>(18)</sup> Venkatswamy. C et al.,<sup>(22)</sup> Rossi A.C et al.,<sup>(6)</sup> and Akgun. H et al.,<sup>(15)</sup>
- During the present study, central nervous system anomalies were seen in 34 out of 150 cases (22.6%). The most common anomalies encountered were meningocele, ventriculomegaly, hydrocephalus and anencephaly. There was a complete agreement between ultrasound and autopsy in 16 out of 34 cases. Additional findings were discovered in 18 cases and in 03 of these cases it led to discovery of syndromes that were initially not suggested in ultrasound. The remaining 15 cases did not change the final diagnosis of ultrasound even after additional findings. The syndromes included anencephaly-spina bifida complex, Joubert syndrome and Hydrolethrus syndrome. It is also noteworthy that ultrasound reports suspected

central nervous system anomalies in 03 cases which could not be confirmed on autopsy. Our findings were directly comparable to findings of Carroll S.G et al.,<sup>(30)</sup> Venkatswamy. C et al.,<sup>(22)</sup> and Nayak S.S et al.,<sup>(18)</sup>

- Skeletal system anomalies were seen in 16 out of 150 cases in our study. A complete agreement between ultrasound and autopsy was seen in 03 cases (23.07%), with additional findings recovered in 13 cases. The most common anomaly observed was congenital talipes equino varus/ valgus. The additional findings led to classification of 07 cases into syndromes. The syndromes included anomalies like complex congenital anomaly, Greenberg dysplasia, VACTERL association and Larsen syndrome. These findings were comparable to a study done by Ceausu,L et al.,<sup>(31)</sup> where they discovered that only 34% of skeletal deformities were discovered during antenatal scans with most common anomaly discovered to be bilateral or unilateral talipes.
- In the study conducted by Vogt. C et al.,<sup>(17)</sup> they observed that a complete agreement between prenatal ultrasound and postmortem findings were established in 84% of the cases. The most common system involved in anomalies was central nervous system, followed by congenital heart defects. This study discovered that minor autopsy findings not seen or recorded at ultrasound examination in 13.4%, major autopsy findings not detected by ultrasound findings 0.4% and none of the autopsy findings suspected in ultrasound examination 0.9%, adding up to a total of 14.7% of additional findings. Struksnaes. C et al.,<sup>(21)</sup> concluded that there was a full agreement between ultrasound and autopsy findings in 88.1% cases, and the main diagnosis was correct in 97.9% cases. In 1.3% cases they could not confirm the ultrasound findings through autopsy. The primary system involved in anomalous fetus was central nervous

system (34.4%), followed by cardiovascular system (18.2%). Additional findings during autopsy were discovered including both minor and major findings in 10.6% of the cases.

- Godbole. K et al.,<sup>(5)</sup> reached to a conclusion that there was complete concordance between ultrasound and autopsy findings in 29.07% cases. Additional information was gathered without influencing the final diagnosis in 46.09% cases and additional information influencing the final diagnosis in 24.82% cases. In the study conducted by Nayak. S.S et al.,<sup>(18)</sup> they observed that antenatal findings could be confirmed in 23% cases with additional findings in 37% cases. In about 23% cases the autopsy changed the final diagnosis and the most common anomaly belonged to central nervous system (13.5%) and then genitourinary system (6.5%). Venkataswamy. C et al.,<sup>(22)</sup> observed a complete agreement in 39.7% cases. Additional findings were noted by them in 62.2% cases and the most common system involved in fetal anomaly was central nervous system (13.6%) which was followed by genitourinary system (6.5%).
- Rossi A.C et al.,<sup>(6)</sup> concluded that the highest proportion of anomalies belonged to central nervous system (36.3%) followed by cardiovascular system (15.6%). They also noted complete concordance between prenatal ultrasound and autopsy findings in 68% cases and discovery of additional findings in autopsy in 22.5% cases<sup>(6)</sup>. Akgun. H et al.,<sup>(15)</sup> observed that 49% cases had central nervous system anomalies, 23% had renal system anomalies and 11 % had congenital heart disease. A complete concordance between ultrasound and autopsy was noted in 77% cases with additional findings seen in 20%. They encountered 3% cases where anomaly was suspected in ultrasound but could not be confirmed in autopsy.
- Results of the present study were comparable to studies done by Vogt. C et al.,<sup>(17)</sup> Struksnaes. C et al.,<sup>(21)</sup> Godbole. K et al.,<sup>(5)</sup> Nayak.S.S et al.,<sup>(18)</sup> Venkatswamy. C et al.,<sup>(22)</sup> Rossi A.C et al.,<sup>(6)</sup> and Akgun. H et al.,<sup>(15)</sup>

- Bhide. P et al.,<sup>(20)</sup> reported that congenital heart defects were the most common anomalies with a prevalence of 65.86 per 10,000 births, followed by skeletal system with prevalence of 49.40 per 10,000 births.
- Unearthing or confirming these skeletal anomalies require additional investigations like performing X-ray. We performed fetal X-rays in 08 cases and in 04 of these cases the X-ray findings aided to the detection of skeletal anomalies like 1) congenital metatarsal varus, congenital dysplasia of humerus and segmentation anomaly of vertebrae along with hypoplasia of vertebral bodies helping in diagnosis of Larsen syndrome (figure 13) 2) short neck, bell shaped thorax, hemivertebrae and thin elongated ribs confirming the diagnosis of complex congenital anomaly 3) butterfly anomaly, segmental anomaly of all lumbar vertebrae with widening and splaying of anterior elements suggestive of lumbar scoliosis (hemivertebrae) thus confirming the presence of VACTERL Association (figure 15) and 4) confirming presence of congenital talipes equino varus/ valgus by showing foot angulation .
- Isaken C.V et al.,<sup>(29)</sup> noted that there was complete agreement between ultrasound and autopsy findings in 70% cases with the most common syndrome being hypoplastic left heart syndrome.

**Table 12: Comparison of present studies with other studies:**

Study	Total sample size	Most common system	Second most common system	Concordance with ultrasound findings	Additional findings
Present study	150	Central nervous system (22.6%)	Skeletal system (10.6%)	54.6%	39.4%
Vogt. C et al., <sup>(17)</sup>	455	Central nervous system (22.9%)	Congenital heart disease (19.7%)	84%	14.7%
Struksnaes. C <sup>(21)</sup>	1029	Central nervous system (34.4%)	Cardiovascular anomalies (18.2%)	88.1%	10.6%
Godbole. K et al., <sup>(5)</sup>	301	Central nervous system (14.9%)	Cardiovascular system (10.2%)	29.07%	70.9%
Nayak. S.S et al., <sup>(18)</sup>	230	Central nervous system	Genitourinary system (6.5%)	23%	37 %

		system (13.5%)			
Venkataswamy. C et al., <sup>(22)</sup>	87	Central nervous system (13.6%)	Genitourinary system (6.5%)	39.7%	62.2%
Rossi A.C et al., <sup>(6)</sup>	3534	Central nervous system (36.3%)	Cardiovascular system (15.6%)	68%	22.5%
Akgun. H et al., (15)	107	Central nervous system (49%)	Renal system (23%)	77%	20%



**Table 13: List of syndromes and associations**

Sr. No.	Syndrome	Findings
1.	Arnold-Chiari malformation (04 cases)	Cyst in posterior fossa, enlarged head, meningocele at lumbosacral region.
2.	Greenberg dysplasia (01 case)	External micromelia, short limbs, thin elongated ribs, hydrops fetalis and cystic hygroma.
3.	Complex congenital anomaly (05 cases)	1) Depressed nasal bridge, generalised scrotal edema, pleural effusion. X-ray: short neck, bell shaped thorax, hemivertebrae and thin elongated ribs. 2) Cystic hygroma, cystic medulla, hypoplastic muscle fibres, dilated lymphatics, single umbilical artery and club foot. 3) Heart apex pointing towards right, part of stomach and left liver lobe seen in hemithorax and edematous external genitalia. 4) Inverted sternal angle, thymic hypoplasia and low set ears 5) Hypertelorism, micrognathia, pulmonary artery right to trachea and thymic aplasia.

4.	Joubert syndrome (01 case)	Bilateral dilated ventricles, thinned out cortex, absent vermis suggestive of hydrocephalus, bilateral low set ears and bilateral congenital equino varus.
5.	Hypoplastic left heart syndrome (02 cases)	Left side of heart is not fully developed, hypoplastic left ventricular wall.
6.	Dandy walker syndrome (02 cases)	Enlarged posterior fossa, hypoplastic or absent cerebellar vermis, cystic dilatation of the fourth ventricle, elevated tentorium cerebelli, possibility of hydrocephalus.
7.	Larsen syndrome (01 case)	Frontal bossing, flattened mid face, hypotelorism, bilateral low set ears, bilateral long and spatulated fingers, bilateral flexed hips and elbows, bilateral hyperextended knees, bilateral club foot underdevelopment of fetal calvarium, fetal sacrum suggestive of hypoplasia, rocker bottom feet and right sided short arm suggestive of congenital skeletal dysplasia. X- ray: congenital metatarsal varus, congenital dysplasia of humerus and segmentation anomaly of vertebrae along with hypoplasia of vertebral bodies.

8.	PAGOD syndrome (01 case)	Bilateral club foot, herniation of stomach in hemithorax, shifting of trachea, dextrocardia with mild left lung hypoplasia.
9.	Anencephaly- Spina bifida complex syndrome (02 cases)	Neural tissue comprised of fibrillary matrix consistent with anencephaly and spina bifida.
10.	Hydrolethalus syndrome (01 case)	Enlarged head, polysyndactyly and cleft lip.
11.	VACTERL association (01 case)	Right toe polydactyly, bilateral CTEV, scoliosis, reduced right ventricular volume and right ventricular hypertrophy.  X- ray: butterfly anomaly, segmental anomaly all lumbar vertebrae with widening and splaying of anterior elements suggestive of lumbar scoliosis.
12.	Pulmonary alveolar proteinosis (01 case)	Distended alveolar spaces with granular eosinophilic PAS positive material.
13.	Harlequinn syndrome (01 case)	Thick, hard skin plates that crack and split, covering the most of their bodies. The plates are diamond shaped and separated by deep fissures.

Assessing the correlation between sonographic results and autopsy findings offers crucial educational insights and feedback for those involved in prenatal diagnosis. It helps identify the areas of limitation and may contribute to enhancing the sensitivity of sonography. Autopsy examination plays a key role in verifying prenatal diagnosis, providing valuable quality control for the sonography team. <sup>(32)</sup>

### **LIMITATIONS OF PRESENT STUDY:**

- Lack of genetic study (karyotyping): The study did not include karyotyping to rule out numerical mutations wherever suspected, restricting our ability to correlate specific genetic mutations with clinical findings.
- Lack of cytogenetic study: The study did not include cytogenetic analysis which could have provided additional insight into abnormalities at molecular level.
- Lack of correlation with placental histopathology: The study did not correlate gross findings with placental morphology, which could have offered more comprehensive understandings of our results and help better analyse spontaneous abortions (intrauterine deaths).

## **SUMMARY:**

- This was a hospital based cross sectional study of 150 fetuses, that included fetuses after spontaneous abortion and medically terminated fetuses.
- Consent was taken from parents in each case. Detailed external examination, gross examination and histopathological examination was done.
- Photodocumentation of significant external and internal developmental anomalies were done.
- Fetal X-rays were performed for 08 cases with indication of skeletal anomalies.
- Complete concordance was noted between sonography and autopsy findings in 54.6% cases with additional findings in 39.4% cases.
- A discordance between ultrasound and autopsy findings was noted in 6% cases.
- Central nervous system was the most involved system followed by skeletal system in our study.
- A total of 23 cases were diagnosed with syndromes and association, which included 12 cases of syndromes that were diagnosed solely based on autopsy.
- The diagnosis can be done on external and gross examination in majority of cases. The histopathological examination played essential role in diagnosis of anomalies like pulmonary alveolar proteinosis and medullary sponge disease.

## **CONCLUSION:**

- The study confirmed the need for fetal pathology examination after fetal loss. The pathologist's contribution to the multidisciplinary management of prenatally diagnosed fetal abnormalities is fundamental for further genetic counselling.
- The study also confirmed the utility of fetal autopsy in identifying the cause of fetal loss, which helps in genetic counselling. In cases with prenatally diagnosed anomalies, the new information obtained from fetal autopsy changes the predicted probability of recurrence risk.
- Even though the prenatal ultrasound reasonably predicts the malformation, fetal autopsy is the gold standard for confirmation of these malformations.

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## ANNEXURE- 1

### INSTITUTIONAL ETHICAL CLEARANCE CERTIFICATE



**BLDE**  
(DEEMED TO BE UNIVERSITY)  
Declared as Deemed to be University u/s 3 of UGC Act, 1956  
Accredited with 'A' Grade by NAAC (Cycle-2)  
The Constituent College

SHRI B. M. PATIL MEDICAL COLLEGE, HOSPITAL & RESEARCH CENTRE, VIJAYAPURA  
BLDE (DU)/IEC/ 939/2023-24 10/4/2023

**INSTITUTIONAL ETHICAL CLEARANCE CERTIFICATE**

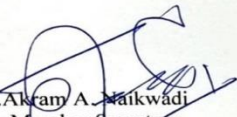
The Ethical Committee of this University met on **Saturday, 18th March, 2023 at 11.30 a.m. in the CAL Laboratory, Dept. of Pharmacology**, scrutinized the Synopsis/ Research Projects of Post Graduate Student / Under Graduate Student /Faculty members of this University /Ph.D. Student College from ethical clearance point of view. After scrutiny, the following original/ corrected and revised version synopsis of the thesis/ research projects has been accorded ethical clearance.

**TITLE: "STUDY OF CORRELATION BETWEEN PRENATAL ULTRASOUND AND MORPHOLOGICAL FINDINGS IN FETAL AUTOPSY AT TERTIARY CARE CENTRE".**

**NAME OF THE STUDENT/PRINCIPAL INVESTIGATOR: DR.AARUSHI GOSWAMI**

**NAME OF THE GUIDE: DR.SAVITRI M. NERUNE , ASSOCIATE PROFESSOR  
DEPT. OF PATHOLOGY.**

Dr. Santoshkumar Jeevangi  
Chairperson  
IEC, BLDE (DU),  
VIJAYAPURA  
**Chairman,  
Institutional Ethical Committee,  
BLDE (Deemed to be University)  
Vijayapura**

  
Dr. Akram A. Maikwadi  
Member Secretary  
IEC, BLDE (DU),  
VIJAYAPURA  
**MEMBER SECRETARY  
Institutional Ethics Committee  
BLDE (Deemed to be University)  
Vijayapura-586103, Karnataka**

Following documents were placed before Ethical Committee for Scrutinization.

- Copy of Synopsis/Research Projects
- Copy of inform consent form
- Any other relevant document

Smt. Bangaramma Sajjan Campus, B. M. Patil Road (Sholapur Road), Vijayapura - 586103, Karnataka, India.  
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## **ANNEXURE -2**

### **CONSENT FOR FETAL AUTOPSY EXAMINATION**

**Date:**

I give my consent to perform a fetal examination (Gross and microscopic examination) on the fetus of \_\_\_\_\_ for which pregnancy was terminated by \_\_\_\_\_ on \_\_\_\_\_. The disposal will be done by the hospital.

I also consent for clinical photographs including the fetus, its internal organs and microscopic examination.

Signature of parents/first-degree relative.

ಭ್ರೂಣದ ಶವಪರೀಕ್ಷೆ ಪರೀಕ್ಷೆಗೆ ಒಪ್ಪಿಗೆ

ದಿನಾಂಕ:

\_\_\_\_\_ ನ ಭ್ರೂಣದ ಮೇಲೆ ಭ್ರೂಣದ ಪರೀಕ್ಷೆಯನ್ನು (ಒಟ್ಟು ಮತ್ತು ಸೂಕ್ಷ್ಮದರ್ಶಕ ಪರೀಕ್ಷೆ)

ಮಾಡಲು ನಾನು ನನ್ನ ಒಪ್ಪಿಗೆಯನ್ನು ನೀಡುತ್ತೇನೆ, ಇದಕ್ಕಾಗಿ \_\_\_\_\_ ರಂದು \_\_\_\_\_

ನಿಂದ ಗರ್ಭಧಾರಣೆಯನ್ನು ಕೊನೆಗೊಳಿಸಲಾಯಿತು. ಭ್ರೂಣದ ಶವಪರೀಕ್ಷೆಯ ನಂತರ ಭ್ರೂಣವನ್ನು

ಆಸ್ಪತ್ರೆಯಿಂದ ವಿಲೇವಾರಿ ಮಾಡಲಾಗುವುದು.

ಭ್ರೂಣ, ಅದರ ಆಂತರಿಕ ಅಂಗಗಳು ಮತ್ತು ಸೂಕ್ಷ್ಮದರ್ಶಕೀಯ ಪರೀಕ್ಷೆ ಸೇರಿದಂತೆ ಕ್ಲಿನಿಕಲ್

ಭಾಯಾಚಿತ್ರಗಳನ್ನು ಪರೀಕ್ಷೆ ಸಹ ನಾನು ಒಪ್ಪಿಗೆಯನ್ನೂ ನೀಡುತ್ತೇನೆ.

ಪೋಷಕರ ಸಹಿ/ಮೊದಲ ಹಂತದ ಸಂಬಂಧಿ.



## **ANNEXURE-3**

### **PROFORMA**

Fetal Autopsy Examination

Name of the patient:

Fetal

autopsy No.

Address/

Tel.

No.:

Consent taken

Date

of

delivery:

Cytogenetics: done/ not done

Fetus

received:

Date,

Time

Referring Doctor

Scan reports: USG

Indication for autopsy

Maternal details

Age

LMP date

Obstetric History

Duration of present pregnancy( gestational age)

H/O consanguineous marriage

Maternal risk factors

Fever

Jaundice

Diabetes mellitus- Blood sugar

Hypertension

Prenatal care

Folic acid supplementation

Pregnancy outcome

Spontaneous abortion

Termination of pregnancy

Intrauterine death

Fetus Measurements:

Weight

Crown Heel Length(CHL)

Crown Rump Length(CRL)

Foot Length

Head Circumference

Inner inter-canthal distance

Chest circumference

Abdominal circumference

Sex

External Examination:

Skin

Head shape

Facial features

Forehead

Eyes- Rt/ Lt

Ears- Rt/Lt

Nose

Mouth

Neck

Umbilicus: 2 vessels/ 3 vessels

External Genitalia:

Male: scrotum/ penis

Female: labia/clitoris

Anus

Patent/ imperforate

Upper Limbs

Forearms

Lower Limbs

Back

In-situ Examination

Cranial Cavity:

Fontanel

Skull Bones

Meninges

Brain

Thoracic cavity

Thymus

Heart- size/situs

Lungs- Bilateral- well developed / collapsed

-Lobes-Rt/ Lt

Diaphragm

Intact/ hernia

Abdomen

Abdominal wall

Situs

Liver

Intestines- appendix

KUB

Internal Examination

Thymus

Weight

Trachea

Esophagus

Heart and Lungs

Weight

Dissection of heart

Liver/ gall bladder

Weight

Spleen

Weight

Pancreas                      Weight

Stomach                      Weight

GIT- small intestines

                    large intestines

Adrenals                      Weight

Kidneys                      Weight

Ureters                      Weight

Urinary Bladder

Internal genitalia

Photographs                      External

                                    Internal

Microscopic Examination:

Autopsy Findings:

## MASTER CHART

S.No	Histopath No.	USG findings	Autopsy findings	Final diagnosis	Additional findings
1.	A/01/24	Arnold chiari syndrome (lemon banana sign, cisterna magna obliterated and lumbar meningocele)	Cyst in posterior fossa, enlarged head, meningocele at lumbosacral region.	Consistent with Arnold chiari syndrome	No additional findings
2.	a/06/24	Normal (Mild pleural effusion)	Normal (dilated alveolar spaces with congested and dilated blood vessels)	Spontaneous abortion	No additional findings
3.	a/07/24	oligohydrominos	Occipital buldge	Spontaneous abortion	Occipital buldge

4.	a/10/24	Dandy walker syndrome (large posterior fossa with vermin hypoplasia, dilated fourth ventricles)	Normal	Could not confirm anomaly	No additional findings
5.	a/11/24	Normal	Normal	Spontaneous abortion	No additional findings
6.	a/12/24	Normal	Normal	Spontaneous abortion	Areas of congestion in heart, liver, spleen and lungs
7.	a/13/24	Normal	Normal	Spontaneous abortion	No additional findings
8.	a/14/24	Normal	Normal	Spontaneous abortion	No additional findings
9.	a/08/24	Normal (monochorionic monoamniotic twin)	Normal	Spontaneous abortion	Areas of congestion in all organs



10.	a/15/24	Bilateral ventriculomegaly, single umbilical artery, thinning of cerebral cortex	Bilateral ventriculomegal y, single umbilical artery, thinning of cerebral cortex	CNS anomaly	No additional findings
11.	a/16/24	Normal	Normal	Spontaneous abortion	Desquamated, squamous and eosinophilic fluid material suggestive of meconium aspiration
12.	a/17/24	Frog eyes suggestive of anencephaly	Absence of cranial spine, occipital area fused with cervical area, absence of cranial vault, spina bifida, webbed neck,	Anencephaly (CNS anomaly)	Absence of cranial spine, occipital area fused with cervical area, absence of cranial vault, spina bifida,

			low set ears, frog eyes		webbed neck, low set ears
13.	a/18/24	CTEV, generalized subcutaneous edema and thick nuchal translucency	Cystic hygroma, cystic medulla, hypoplastic muscle fibres, dilated lymphatics, SUA, CTEV	Complex Congenital Anomaly	cystic medulla, hypoplastic muscle fibres,
14.	a/19/24	Normal  (subchorionic bleed and low lying placenta)	Normal	Spontaneous abortion	No additional findings
15.	a/20/24	Agenesis of corpus callosum	Agenesis of corpus callosum	CNS anomaly	No additional findings
16.	a/21/214	Hydrops fetalis and skeletal dysplasia	Depressed nasal bridge, generalized scrotal edema suggestive of	Complex Congenital Anomaly	X-Ray findings- short neck, bell shaped thorax, hemivertebrae and thin

			hydrops fetalis, pleural effusion.  X- Ray findings- short neck, bell shaped thorax, hemivertebrae and thin elongated ribs		elongated ribs, pleural effusion
17.	a/23/24	Slopping frontal lobe and persistent SVC	Slopping frontal lobe and persistent SVC	CVS anomaly	No additional findings
18.	a/09/24	Normal  (subchorionic bleed and low lying placenta)	Normal	Spontaneous abortion	No additional findings
19.	a/24/24	Hemivertebrae and bilateral pyelectasis	Hemivertebrae, contractures in bilateral upper and lower limb	Renal anomaly	contractures in bilateral upper and lower limb
20.	a/25/24	Skeletal dysplasia and cystic hygroma	Micromelia, short limbs, thin	Greenberg Dysplasia	Micromelia, short limbs,

			elongated ribs, hydrops fetalis and cystic hygroma	(Skeletal anomaly)	thin elongated ribs, hydrops fetalis
21.	a/26/24	Mild ventriculomegaly and skeletal dysplasia	Dilated ventricles and shortened upper and lower limbs	CNS anomaly	No additional findings
22.	a/27/24	Differential diagnosis of Dandy walker syndrome and Joubert syndrome	Thin brain parenchyma, bilateral dilated ventricles, cystic dilatation suggestive of Dandy walker syndrome enlarged head and protruding eyes	Dandy walker syndrome  (CNS anomaly)	Thin brain parenchyma, bilateral dilated ventricles, cystic dilatation
23.	a/28/24	Bilateral echogenic kidneys	Placenta showing hydropic	Skeletal anomaly	Placenta showing hydropic

			degeneration, low set ears, webbed neck and polydactyly (all four limbs)		degeneration, low set ears, webbed neck and polydactyly
24.	a/29/24	Normal	Normal (coiled umbilical artery and SUA)	Spontaneous abortion	coiled umbilical artery and SUA
25.	a/30/24	HLHS	Conical shaped heart, hypoplastic left wall and bilateral hip joint ulceration	HLHS (CVS anomaly)	bilateral hip joint ulceration
26.	a/31/24	Normal (anhydrominos)	Normal	Spontaneous abortion	No additional findings
27.	a/32/24	Dextrocardia, stomach, part of small bowel and left liver lobe seen in hemithorax	Dextrocardia, stomach, part of small bowel and left liver lobe seen in	Complex congenital anomaly	Edematous external genitalia

			hemithorax and edematous external genitalia		
28.	A/33/24	Normal	Normal	Spontaneous abortion	No additional findings
29.	a/34/24	Normal	Normal (orbital edema)	Spontaneous abortion	orbital edema
30.	a/39/24	Dandy walker syndrome, skeletal dysplasia and CTEV	Cystic posterior fossa suggestive of dandy walker syndrome, club foot suggestive of skeletal dysplasia and enlarged head	Complex congenital anomaly	enlarged head
31.	a/40/24	Normal	Normal	Spontaneous abortion	No additional findings
32.	a/42/24	Lemon and banana sign, meningomyelocele	Head appears elongated, meningocele at	CNS anomaly	No additional findings

			lumbosacral region		
33.	a/43/24	Normal	Normal	Spontaneous abortion	No additional findings
34.	a/45/24	Normal	Normal (low set ears)	Spontaneous abortion	Low set ears
35.	a/46/24	Normal  (retroplacental hematoma)	Normal	Spontaneous abortion	No additional findings
36.	a/47/24	Normal	Normal  (greenish discolouration of mouth and SUA)	Spontaneous abortion	SUA
37.	a/48/24	Larsen syndrome- decreased intraorbital distance, hypoplastic face, inward left foot angulation, CTEV, prefrontal edema, genu recurvatum	Frontal bossing, flattened mid face, hypotelorism, bilateral low set ears, bilateral long and	Larsen syndrome	X-ray: congenital metatarsal varus, congenital dysplasia of humerus and

			spatulated fingers, bilateral flexed hips and elbows, bilateral hyperextended knees, bilateral club foot underdevelopment of fetal calvarium, fetal sacrum suggestive of hypoplasia, rocker bottom feet and right sided short arm suggestive of congenital skeletal dysplasia. X- ray: congenital metatarsal varus, congenital		segmentation anomaly of vertebrae along with hypoplasia of vertebral bodies.
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			dysplasia of humerus and segmentation anomaly of vertebrae along with hypoplasia of vertebral bodies.		
38.	a/49/24	Twin A- absent diastolic flow  Twin B- cardiomegaly with ventricular wall hypertrophy	Twin A- absent diastolic flow and SUA  Twin B- cardiomegaly with ventricular wall hypertrophy	Twin to twin transfusion syndrome	Twin A- SUA
39.	a/50/24	Normal	Normal	Spontaneous abortion	No additional findings
40.	a/51/24	Left lung diaphragmatic hernia and left lung hypoplasia	Bilateral club foot, herniation of stomach into	PAGOD syndrome	Bilateral club foot, shifting of trachea,

			hemithorax, shifting of trachea, dextrocardia, mild lung hypoplasia, scrotal edema		dextrocardia and scrotal edema
41.	a/53/24	Normal (anhydrominos)	Normal (left eyelid everted)	Spontaneous abortion	left eyelid everted
42.	a/54/24	Right ectopic kidney with hydrouretronephrosis	Right kidney agenesis and SUA	Renal anomaly	SUA
43.	a/56/24	Normal (retroplacental clot)	Normal (congestion of bilateral lungs)	Spontaneous abortion	congestion of bilateral lungs
44.	a/55/24	IUGR	Decreased stature	Skeletal system	No additional findings
45.	a/57/24	Unilateral ventriculomegaly	Normal	Could not confirm USG findings	No additional findings

46.	a/60/24	Normal	Normal (flat nasal bridge and low set ears)	Spontaneous abortion	flat nasal bridge and low set ears
47.	a/62/24	Normal	Normal	Spontaneous abortion	No additional findings
48.	a/63/24	Increased nuchal translucency, kyphoscoliosis, spinal dysmorphism and diastematomyelia	kyphoscoliosis, spinal dysmorphism and subcutaneous defect in lumbar region	CNS anomaly	No additional findings
49.	a/65/24	Normal	Normal (low set ears)	Spontaneous abortion	low set ears
50.	a/66/24	Normal	Normal	Spontaneous abortion	No additional findings
51.	a/67/24	Frog facies (anencephaly)	Anencephaly, shoulder dystocia and imperforate anus	CNS anomaly	shoulder dystocia and imperforate anus

52.	a/98/24	Normal	Normal (cephalhematom a)	Spontaneous abortion	cephalhemato ma
53.	a/69/24	Normal	Normal	Spontaneous abortion	No additional findings
54.	a/70/24	Gastrochisis (colonic atresia)	Herniated bowel loops through anterior abdominal wall suggestive of gastrochisis and low set ears	GIT anomaly	low set ears
55.	a/73/24	Normal (oligohydrominos and IUGR)	Normal (SUA)	Could not confirm IUGR	SUA
56.	a/76/24	Normal	Normal (bilateral lung congestion)	Spontaneous abortion	bilateral lung congestion

57.	a/76/24	Normal	Normal (bilateral lung congestion)	Spontaneous abortion	bilateral lung congestion
58.	a/32/23	Normal (low lying placenta)	Normal	Spontaneous abortion	No additional findings
59.	a/33/23	Normal (low lying placenta)	Normal	Spontaneous abortion	No additional findings
60.	a/34/23	Normal	Normal (flat head)	Spontaneous abortion	flat head
61.	a/35/23	Spina bifida	Normal	Could not confirm grossly	No additional findings
62.	a/41/23	Anencephaly complex anomaly	Neural tube comprised of fibrillary matrix consistent with anencephaly and spina bifida	Anencephaly - spina bifida complex	No additional findings

63.	a/42/23	Normal	depressed skull and right kidney tubular necrosis	Renal anomaly	depressed skull and right kidney tubular necrosis
64.	a/44/23	Normal	Normal	Spontaneous abortion	No additional findings
65.	a/45/23	Normal(abruptio placenta)	Normal (congested organs)	Spontaneous abortion	Congested organs
66.	a/46/23	Dysplastic mitral valves and hypoplastic left heart	Normal	Could not confirm USG findings	No additional findings
67.	a/47/23	Right mild pelviectasis and left diaphragmatic hernia	left diaphragmatic hernia (both kidneys normal)	Could not confirm renal anomaly	No additional findings
68.	a/48/23	Normal(oligohydromin os)	Normal	Spontaneous abortion	No additional findings
69.	a/49/23	Normal(abruptio placenta)	Normal	Spontaneous abortion	No additional findings

70.	a/50/23	Tetralogy of fallot and single umbilical artery	Ventricular septal defect, muscle hypertrophy and variable size cardiomyocytes (Tetralogy of fallot) and SUA	CVS anomaly	Cardiac muscle hypertrophy
71.	a/53/23	Hydrocephalus, polysyndactyly and cleft lip	Enlarged head, polysyndactyly and cleft lip	Hydroletharus syndrome	No additional findings
72.	a/54/23	Occipital encephalocele	Small head	CNS anomaly	No additional findings
73.	a/55/23	Normal(oligohydrominos)	Normal (congested organs)	Spontaneous abortion	No additional findings
74.	a/92/23	Invertus siatus, levocardia and right aortic arch	Invertus siatus, levocardia, hypoplasia of thymus, low set	Complex congenital anomaly	Hypoplasia of thymus, low set ears

			ears and right aortic arch		
75.	a/57/23	Normal(oligohydrominos)	Normal (congested organs)	Spontaneous abortion	No additional findings
76.	a/58/23	Normal	Normal (occipital protruberance)	Spontaneous abortion	occipital protruberance
77.	a/59/23	AVSD	Dilated SVC, double outlet right ventricle, low set ears	CVS anomaly	low set ears
78.	a/60/23	Bilateral hydrocephalus	Dilatation of posterior horn, moderate ventriculomegal y and globular head	CNS anomaly	globular head
79.	a/62/23	Normal	Normal (	Spontaneous abortion	No additional findings



80.	a/63/23	Hypertelorism, micrognathia, pulmonary artery right to trachea, left pulmonary artery anterior to trachea	Hypertelorism, micrognathia, pulmonary artery right to trachea, left pulmonary artery anterior to trachea and thymus hypoplasia	Complex congenital anomaly	Thymus hypoplasia
81.	a/68/23	Normal	Normal	Spontaneous abortion	No additional findings
82.	a/65/23	Normal	Normal	Spontaneous abortion	No additional findings
83.	a/66/23	Arnold chiari syndrome	Meningocele, left sponge kidney and cystic kidney	CNS and Renal system	left sponge kidney and cystic kidney
84.	a/69/23	Normal	Normal	Spontaneous abortion	No additional findings

85.	a/70/23	Anencephaly	Anencephaly, low set ears	CNS anomaly	Low set ears
86.	a/71/23	Arnold chiari syndrome and left foot tallipes	Left foot tallipes	Could not confirm CNS anomaly	No additional findings
87.	a/72/23	Normal (anhydrominos)	Normal	Spontaneous abortion	No additional findings
88.	a/73/23	Normal	Normal	Spontaneous abortion	No additional findings
89.	a/75/23	Normal (oligohydrominos)	Normal	Spontaneous abortion	No additional findings
90.	a/76/23	Normal (anhydrominos)	Normal	Spontaneous abortion	No additional findings
91.	a/77/23	Normal (oligohydrominos)	Normal	Spontaneous abortion	No additional findings
92.	a/78/23	Increased nuchal translucency, hypoplastic nasal bone	Right ventricular hypertrophy, pulmonary stenosis, boot	CVS anomaly	Right ventricular hypertrophy, pulmonary stenosis, boot

			shaped heart and low set ears		shaped heart and low set ears
93.	a/79/23	Bilateral dilated ventricles	Spinal defect comprised of skin, meningeal layers and neural tissue along with low set ears	CNS anomaly	Spinal defect comprised of skin, meningeal layers and neural tissue along with low set ears
94.	a/80/23	Anencephaly	Anencephaly, protruding tongue and absence of nasal bone	CNS anomaly	protruding tongue and absence of nasal bone
95.	a/81/23	Cystic hygroma	Cystic hygroma	Lymphatics anomaly	No additional findings
96.	a/82/23	Ventricular septal defect, dilated aorta and unossified nasal bone	Ascending aorta and aortic arch dilated, inlet VSD	CVS anomaly(could not confirm)	No additional findings

				other CVS findings)	
97.	a/83/23	Tetralogy of falot	Right toe polydactyly, bilateral CTEV, scoliosis, reduced right ventricular volume and right ventricular hypertrophy.  X- ray: butterfly anomaly, segmental anomaly all lumbar vertebrae with widening and splaying of anterior elements suggestive of lumbar scoliosis.	VACTERL Association	Right toe polydactyly, bilateral CTEV, scoliosis, reduced right ventricular volume and right ventricular hypertrophy.  X- ray: butterfly anomaly, segmental anomaly all lumbar vertebrae with widening and splaying of anterior

					elements suggestive of lumbar scoliosis.
98.	a/84/23	Normal (anhydrominos)	Normal (periorbital edema)	Spontaneous abortion	periorbital edema
99.	a/85/23	Normal	Normal (low set ears)	Spontaneous abortion	low set ears
100.	a/86/23	Normal (oligohydrominos)	Normal	Spontaneous abortion	No additional findings
101.	a/87/23	Normal (retroplacental clot)	Normal	Spontaneous abortion	No additional findings
102.	a/90/23	Ectopic kidney, hemivertebrae	Ectopic kidney within intestinal loops, hemivertebrae and thoracic scoliosis	CNS and Renal system anomaly	thoracic scoliosis

103.	a/88/23	Normal	Normal	Spontaneous abortion	No additional findings
104.	a/91/23	Cleft lip and palate	Cleft lip and palate	Spontaneous abortion(Facial anomaly)	No additional findings
105.	a/113/24	Normal (severe oligohydrominos and abruptio placenta)	Normal (congested and dilated alveolar spaces)	Spontaneous abortion	congested and dilated alveolar spaces
106.	a/121/24	Bilateral CTEV, hydrocephalus	Bilateral dilated ventricles, thinned out cortex, absent vermis suggestive of hydrocephalus, bilateral low set ears and bilateral congenital equino varus.	Joubert syndrome	bilateral low set ears

107.	a/128/24	Subcutaneous edema and frontal bossing	Normal	Could not confirm USG findings	No additional findings
108.	a/130/24	Absent ductus venosus, cystic hygroma, bilateral minimal pleural effusion suggestive of nonimmune hydrops fetalis and low lying placenta	Ruptured head and abdomen with visible intestinal loops and cystic hygroma	GIT anomaly	No additional findings
109.	a/133/24	Well defined cystic lesion in posterior fossa suggestive of blake pouch cyst	Partially opened up alveoli and congestion, yellow fluid drained	Blake pouch cyst	No additional findings
110.	a/134/24	Lemon shaped frontal bones, prominent ventricles and banana sign (Arnold chiari malformation)	Frog eyes, lemon head and low set ears	Arnold chiari malformation	Low set ears

111.	a/140/24	Absent skull bone, facial dysmorphism suggestive of anencephaly and low lying placenta	Absent skull bone, no forehead and no mouth, no eyes	Anencephaly	No additional findings
112.	a/145/24	Uteroplacental insufficiency, IUGR	Distended alveolar spaces with PAS positive eosinophilic granular material	Pulmonary alveolar proteinosis	Distended alveolar spaces with PAS positive eosinophilic granular material
113.	a/149/24	Normal (anhydrominos)	Normal	Spontaneous abortion	No additional findings
114.	a/151/24	Severe ventriculomegaly and SUA	Severe ventriculomegaly and SUA	CNS anomaly	No additional findings
115.	a/144/24	Normal	Normal	Spontaneous abortion	No additional findings



116.	a/143/24	oligohydrominos, mild moulding of skull bones	low set ears and tiny skull	CNS anomaly	low set ears
117.	a/142/24	Normal	Normal	Spontaneous abortion	No additional findings
118.	a/141/24	Normal	Normal	Spontaneous abortion	No additional findings
119.	a/136/24	Normal	Normal (low set ears)	Spontaneous abortion	Low set ears
120.	a/135/24	Normal	Normal (low set ears)	Spontaneous abortion	Low set ears
121.	a/132/24	Normal (polyhydrominos)	Normal (underdeveloped ears)	Spontaneous abortion	Underdeveloped ears
122.	a/131/24	Absent diastolic flow suggestive of growth restriction	Normal	Could not confirm USG findings	No additional findings
123.	a/129/24	Twin A- retained products of conception	Imperforate anus and left foot four fingers	Skeletal anomaly	Imperforate anus and left

					foot four fingers
124.	a/129/24	Twin B- Retained products of conception	Underdeveloped baby, right lung two lobes arteries and veins not identified	Twin to twin transfusion syndrome	Underdeveloped baby, right lung two lobes arteries and veins not identified
125.	a/124/24	Normal	Normal	Spontaneous abortion	No additional findings
126.	a/123/24	Normal	Globular head	Spontaneous abortion	Globular head
127.	a/122/24	Severe oligohydrominos and bradycardia	Enlarged head and webbed neck	CNS anomaly	Enlarged head and webbed neck
128.	a/120/24	CTEV	Globular head, facial features distorted from nose to chin, bilaterally medially	Skeletal anomaly	Globular head, facial features distorted from nose to chin,

			deviated foot and foot appears planta flexed suggestive of CTEV		
129.	a/119/24	Normal (uteroplacental insufficiency)	Normal	Spontaneous abortion	No additional findings
130.	a/117/24	Normal	Normal	Spontaneous abortion	No additional findings
131.	a/116/24	Normal	Low set ears, cleft lip and palatae	Facial anomaly	Low set eras, cleft lip and palate
132.	a/115/24	Normal	Normal	Spontaneous abortion	No additional findings
133.	a/118/24	Normal	Normal	Spontaneous abortion	No additional findings
134.	a/146/24	Normal	Normal	Spontaneous abortion	No additional findings

135.	a/147/24	Normal	Normal	Spontaneous abortion	No additional findings
136.	a/148/24	Normal	Normal (low set ears)	Spontaneous abortion	Low set ears
137.	a/71/24	Severe oligohydrominos	Webbed neck and enlarged head	CNS anomaly	Webbed neck and enlarged head
138.	a/64/24	Anencephaly	Anencephaly, low set ears, SUA	CNS anomaly	Low set ears and SUA
139.	a/72/24	Normal	Normal	Spontaneous abortion	No additional findings
140.	a/68/24	HLHS	Conical heart, hypoplastic left wall, SUA	HLHS	SUA
141.	a/130/24	Normal	Normal	Spontaneous abortion	No additional findings

142.	a/150/24	Normal	Normal (congested organs)	Spontaneous abortion	Congested organs
143.	a/151/24	Anencephaly complex anomaly	Neural tissue comprised of fibrillary matrix consistent with anencephaly along with spina bifida	Anencephaly - spina bifida complex	No additional findings
144.	a/155/24	Harlequinn syndrome	Thick, hard skin plates that crack and split, covering the most of their bodies. The plates are diamond shaped and separated by deep fissures.	Harlequinn syndrome	No additional findings

145.	a/36/23	Normal	Normal	Spontaneous abortion	No additional findings
146.	a/37/23	Normal (abruptio placenta)	Normal (congested organs)	Spontaneous abortion	Congested organs
147.	a/38/23	Normal	Normal	Spontaneous abortion	No additional findings
148.	a/39/23	Retained products of conception	Normal (Low set ears)	Spontaneous abortion	Low set ears
149.	a/40/23	Normal (retroplacental hematoma)	Normal	Spontaneous abortion	No additional findings
150.	a/43/23	Lemon and banana sign, meningomyelocele	Head appears elongated, meningocele and lumbosacral region	CNS anomaly	No additional findings

# aarushi goswami

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