

## Study on various congenital anomalies in fetal autopsy

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### Abstract

**Introduction:** The study of dead is to save the livings. Congenital malformations have become important cause of fetal and neonatal (perinatal) mortality in developed countries and would very soon be increasingly important determinants of fetal mortality in developing countries like India. The fetal autopsy plays the vital role in the conformation, identification of congenital anomalies and also for the counselling of the parents, to prevent the fetal congenital anomalies in further pregnancies. This study was undertaken with the purpose of finding out cause of death during the perinatal period at government kilpauk medical college Chennai and to study the clinical and pathological findings in fetal death. **Methods:** The retrospective study of congenital anomalies in fetal deaths was done at government kilpauk medical college Chennai over a time period of 1 years from June 2015 to June 2016. The present study included dead fetus. autopsy was performed by standard technique. External and internal findings are followed by histopathological examination and autopsy findings were compared with available ultrasound findings. **Results:** A total of 7 Autopsies performed. , there were 4 male and 3 female babies. Two babies showed external malformation and five babies showed internal congenital anomalies. Congenital anomalies were commonest in the birth weight group of 100-800 grams. Malformations of central nervous system, cardiovascular system and abdomen were most common followed by genitourinary and musculoskeletal system respectively. **Conclusion:** Perinatal deaths occurred in low birth weight and preterm babies. Study of malformations greatly helpful in genetic counselling and prenatal diagnosis in successive pregnancies.

**Keywords:** Fetal, Autopsy, Congenital anomalies

### Introduction

The incidence of congenital anomalies has been increasing in recent past. The worldwide incidence of congenital disorder is estimated at 3-7%, but actual numbers vary widely due to under reporting of cases in developing countries [1]. The presence of congenital abnormalities in a baby has an emotional effect not only on the mother but also on the family [2]. A congenital anomaly is defined in terms of physical structure as a malformation, an abnormality of physical structure or form usually found at birth or during the first few weeks of life [3]. Congenital malformations remain a common cause of perinatal deaths accounting for 10-15% in developing countries like India [4]. Unlike the situations in developed countries, where congenital

malformations are leading cause of infant mortality, in India low birth weight, prematurity, sepsis and infections are still the leading causes. Perhaps for this reason not much attention has been paid to the problem of congenital malformations in India [5]. In spite of antenatal diagnostic modality still the fetal autopsy plays the vital role in the conformation as well as identification of congenital anomalies and also for the counselling of the parents, to prevent the fetal congenital anomalies in further pregnancies. The fetal and neonatal (perinatal) mortality serves as the most sensitive index of maternal and neonatal care. In an area it also reflects the general public health and socio biological features of mothers and infants. The incidence of the perinatal mortality differs between different countries. Simultaneously repeated evaluation of this problem is very essential to study their magnitude and

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causative factor which shows the impact of existing medical facilities as well as need for their expansion [6]. Why do we examine the aborted fetus?, the primary reason must be to device information which can be used to counsel parents in respect of future pregnancies either in the context of recurrence risk of fetal malformations, a subsequent spontaneous abortion (or) intrauterine fetal death.

Additional important factors in an audit functions in respect of prenatal diagnosis of malformations. It also provides an opportunity to examine the malformed fetus at a stage earlier in development than was possible previously. Such examinations may contribute to our understanding of the processes of disordered fetal development, this in turn improve treatment of affected live born infant. With the increasing use of ultrasonography for prenatal diagnosis, it has become particularly important to examine the fetus following termination of pregnancy for anomaly. Ultrasound examination is a very effective detector of major structural anomalies, but lacks the specificity which is essential for syndrome diagnosis. Thus fetal examination has a largely confirmatory role when termination of pregnancy has been under taken because of prenatally detected chromosome anomaly (or) genetic metabolic disease, structural anomalies may be present.

When invasive procedures have been under taken for diagnosis or therapy, fetal examination should include a deliberate search for traumatic result. All aborted fetus should be transported rapidly to the pathology department, in a fresh, unfixated state. They should be accompanied by appropriate clinical information, so that all essential investigations can be under taken with due dispatch. The recurrence risk of these anomalies varies from negligibly low to 25% depending on the genetic component in the etiology of the disorder.

## Methods

The present study is retrospective study of congenital anomalies in fetal deaths was done at Government kilpauk medical college, Chennai. Study conducted over a time period of 1 year from June 2015 to June 2016. The dissecting instruments required for fetal and neonatal (perinatal) autopsy are small scissors and forceps and scalpels. The autopsy protocol was including scales for recording specific measurements and norms for particular gestational ages.

Measurements: The Crown Heel (CH) and Crown Rump (CR) lengths should be determined to the nearest 0.5 cum. Chest and abdominal circumferences were taken at the level of the nipple and umbilicus respectively. Foot length correlates especially well with gestational age, and should be obtained in every case. The metric documentation of changes in the face is often a valuable component of the autopsy. The distances between the inner canthi and outer canthi, nasal height and width, philtrum height, mouth width and ear length can be obtained and compared with

Every effort should be made to identify the etiology of the perinatal death so that appropriate genetic counselling can be given [7]. When any general study of still born is undertaken it is highly desirable that these deaths are clarified as to the cause.

A perinatal autopsy can provide an explanation for a loss often relieving the patient and her physician of blame and may reveal a specific disorder for which precise recurrence risk (or) strategies for prevention are available perinatal mortality is going important due to the fact that maximum loss of human life occurs in that period. Such loss is often difficult for patients and their families [8]. The autopsy plays a critical and multifaceted role in modern medicine not the least of which is its function as a quality control and verification mechanism in diagnosis with its ultimate salutary impact on clinical practice [9].

Autopsy studies are important and essential in new born death as the clinical findings are confusing and in spite of thorough pathological investigations, it may not be possible to arrive at the complete pathological diagnosis. This study was undertaken with the purpose of finding out cause of death during the perinatal period at Government kilpauk medical college, Chennai for the period of from June 2015 to June 2016.

## Aims and objectives

1. To study of the various lesions (congenital anomalies) in fetal Autopsy.
2. To assess the predisposing factors and cause of death by the autopsy method.
3. To study the clinical and pathological findings (Gross & microscopic) in fetal death.
4. To take necessary preventive aspects (counselling) of congenital anomalies.

published norms. Weights: Scales accurate to 0.1 gm. are necessary for perinatal specimens. All major organs should be weighed (i.e. thymus, heart, lungs, liver, spleen, kidney, adrenal glands, brain and placenta) and the date recorded in the autopsy protocol along with expected values. Photographs were taken, they provide indisputable evidence of findings, sharing of dysmorphic face images to geneticist was done for important diagnostic information.

The present study included all dead fetus and Autopsy was performed by standard technique adopted by Edith L. Potter. External examination done for inspection of cyanosis, injuries and maceration, skin lesions, all major and minor developmental anomalies were described. The Y shaped incision was done which extend from the anterior aspect of each shoulder to the xiphoid process. Umbilical vein should be examined for signs of inflammation, varix, rupture (or) thrombus. The two umbilical arteries are examined and inspected in their entirety. The arteries and urachus should be examined for patency and the arteries for haemorrhage (or) thrombosis. Single umbilical artery was an important anomaly and should be documented.

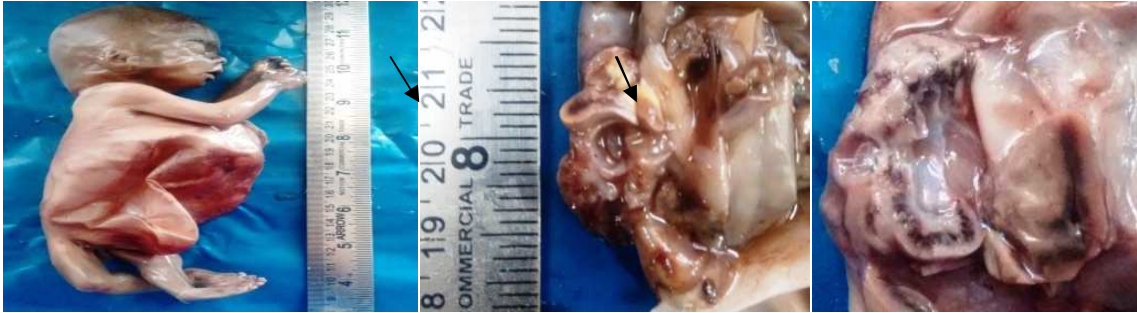
The autopsy protocol included the removal of thoracic, cervical, abdominal and pelvic organs en bloc and subsequently dissected into organ blocks. Internal examination: All internal organs position and size were examined. The internal genitalia are inspected. The testis will be undescended in younger foetuses and removed with abdominal contents. Prior opening the pleural cavities the possibility of pneumo thorax should be entertained. Upon entering the chest each cavity should be inspected for fluid, each lung was examined for developmental changes carefully. The integrity and tension of the pericardium are ascertained and the pericardial cavity opened again, the presence of free gas (or) fluid is determined. Any fibrinous deposition over the surface of pericardium (pericarditis) sent for culture or microscopy. Heart is best examined in situ, while anatomic relationship with structures was intact, then inspected externally and internally, in a systematic fashion that follows the movement of the blood. All major veins and arteries were examined.

The diagnosis of premature closure of foramen ovale can be made. The configuration of tricuspid valve, right ventricle, and main pulmonary artery were studied. The endocardium, myocardium, and configuration of trabeculae, pectinate and papillary muscles and chordae tendineae were examined. After opening the left heart, the interior of the left atrium, pulmonary venous orifices, mitral valve and left ventricle were inspected, followed by examination of the aortic valve and ascending aorta. All other organs were removed as en bloc. Neck structures trachea and esophagus were examined.

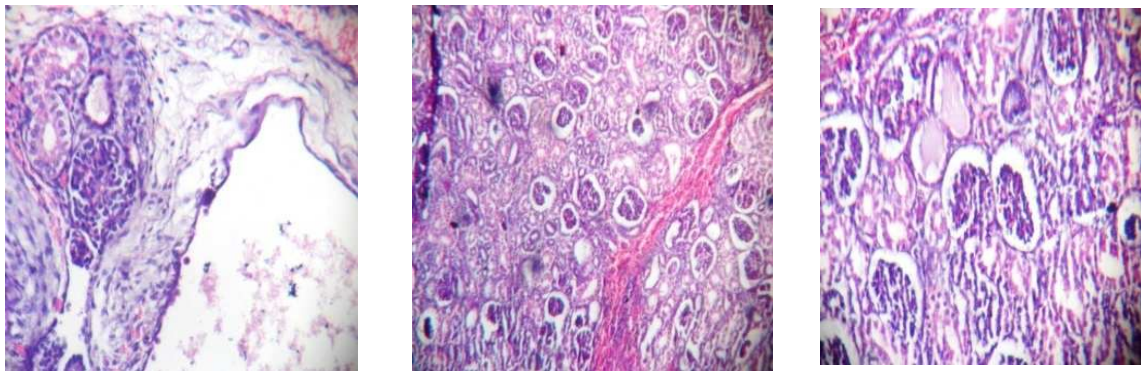
The scalp, fontanel, and cranial sutures were examined by palpation and any changes were documented. The fontanel, sutures, and glia were examined and any changes were documented. After brain has been exposed, it was examined in situ. After the brain was removed, it is examined on all sides and placed in fixative. Attention should then be turned to the cranial base and dural sinuses. The pituitary gland is removed from the sella turcica by careful dissection.

Dissecting the viscera: The Rokitansky methods of evisceration, the organs were removed together in a blocks [10]. Examination begins with the most posterior structures and moves anteriorly layer by layer. Aorta, inferior vena cava, adrenal glands and posterior surface of the urinary system exposed and examined. Adrenal glands, kidneys, ureters and urinary bladder were examined. The vagina and uterus were opened in the anterior midline and examined. The liver, gallbladder and structures of the porta hepatis, portal vein, hepatic artery and common bile duct were identified and dissected as indicated. The esophagus was opened in the posterior midline while intact with the trachea. In this way a tracheoesophageal fistula can be identified. After opening, the incision may be carried into the stomach. After major hilar structures of the lungs have been opened and inspected, attention was turned to the lungs themselves. Lobation and condition of the visceral pleura were presumably ascertained. In case of bladder outlet obstruction, the entire urethra must be examined for posterior urethral valves (or) other abnormalities (i.e., anterior urethral valves, mega urethra). Placenta was available in only few cases.

Histopathological examination: The organs after evisceration and external examination were fixed in 10% formalin. Blocks of tissues for microscopic examination were taken, one block from each lobe of both lungs. One block each from thymus, heart, stomach, liver, spleen, pancreas, small intestine, large intestine, kidneys, adrenals, and any doubtful lesions were taken. Sections were studied in the routine way with Haematoxylin and Eosin (H & E) stains. Autopsy findings were compared with ultrasound findings whenever available.



**Image-1: Gross- 20wks gestation fetus with multiple cyst in kidney**



**Image-2: Histology of multicyst with dystrophic tubules**



**Image-3: Gross – 22 Wks gestation with b/l ventriculomegaly**



**Image-4: 22wks gestation with meningocele**



**Image-5: 24 Wks Gestation with Musculoskeletal Defect**



**Image-6: 21wks gestation with congenital diaphragmatic hernia & dextrocardia**

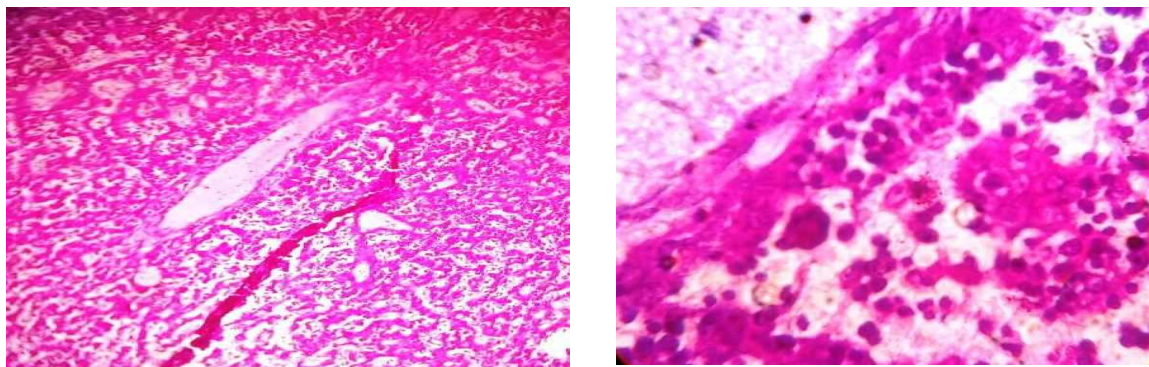


**Image-7: 14 wks gestation with abdominal wall defect**



**Image-8: 22wks Gestation with Hepatomegaly**





**Image-9: Histology of liver showing extramedullary haematopoiesis in low & high power**

**Results**

A total of 7 Autopsies performed. In a total of 7 foetuses, there were 4 male and 3 female babies. On external examination of 7 fetal 3 babies showed congenital malformation. On internal examination of 7 fetus, 4 babies showed internal congenital anomalies. Congenital anomalies were commonest in the birth weight group of 100-800 grams accounting for 7 cases. Malformations of central nervous system, cardiovascular system and abdomen were most common followed by genitourinary and musculoskeletal system respectively. The age of mothers of these babies ranged from 19 years to 30 years. Maternal age above 35 years was not found to be a significant contributing factor towards unexplained fetal death [11]. Of the 7 fetal death, 2 babies were born to primi gravida followed by 4 babies who were born to gravida-II. 1 baby was born to gravida->III. The gestational age of 7 foetuses ranged from 14 weeks to 24 weeks. Of total 7 fetal death the weight of 5 foetuses were <500 gm and weight of 2 foetuses were between 500-1000 gm.

External congenital anomalies: On external examination of 7 fetal deaths, 3 babies showed congenital malformation, (Table 1).

**Table-1: External congenital anomalies.**

System affected	Type of anomaly	No	Total
CNS	Meningomyelocele	1	1
Abdomen	Abdomen wall defect	1	1
Musculoskeletal	Absence of limb,digit	1	1
<b>Total</b>		<b>3</b>	<b>3</b>

Internal anomalies: On internal examination of the 7 fetal deaths, 4 babies showed internal congenital anomalies, (Table 2).

**Table-2: Internal congenital anomalies.**

System affected	Type of anomaly	No	Total
Renal	Multicystic kidney disease	1	1
Heart	Ventriculomegaly	1	1
Thorax	Congenital diaphragmatic hernia	1	1
Abdomen	Hepatomegaly	1	1
<b>Total</b>		<b>4</b>	<b>4</b>

Out of 7 fetal death 2 babies are primigravida and 5 babies are multigravida.all fetus have congenital anomaly Fisher P value <.05). 71.4% of fetus with weight less than 500 gm and 28.57% of fetus with weight range b/w 500-1000 gm. have congenital anomalies and all congenital anomalies have birth weight less than 1000 gm. (Fisher Exact P value <.05). All cases are 100% correlated with ultra sonogram.

## Discussion

Fetal autopsy significantly contributes to the diagnosis of intrauterine fetal death and congenital anomalies are a major cause of perinatal death [11]. Congenital malformations in fetal and neonatal deaths vary in different studies. Musculoskeletal, cutaneous and genitourinary malformations were common among live born babies while central nervous system and gastrointestinal defects were common among still born babies [12].

The study of malformations greatly helpful in genetic counselling and prenatal diagnosis in successive pregnancies. In the present study 7 fetal autopsy was carried out at government kilpauk medical college, Chennai.

In our study male and female ratio is 1.3: 1. All fetus are expelled by MTP at 20-24 wks and the birth weight was between 100-800gms. The most common anomalies were related to the cardiac and abdominal anomaly followed by renal, musculoskeletal and CNS. The CNS anomalies occur due to defective closure of Neural tube between the 23rd and 26th day of gestation resulting in anencephaly or meningomyelocele (image 4). This study coincided with the study of, P. A. Boyd et al. (2003) [15].

	CASE 1	CASE 2	CASE 3	CASE 4	CASE 5	CASE 6	CASE 7
<b>Maternal age</b>	22	23	20	30	24	19	22
<b>Gravida</b>	Primi	2 <sup>nd</sup> gravid	Primi	6 <sup>th</sup> gravid	2 <sup>nd</sup> gravid	2 <sup>nd</sup> gravid	3 <sup>rd</sup> gravida
<b>Consanguinity</b>	3 <sup>rd</sup> degree	Absent	Absent	2 <sup>nd</sup> degree	2 <sup>nd</sup> degree	Absent	Absent
<b>Gestational age</b>	20	20	22	24	21	14	22
<b>Correlation with USG</b>	+	+	+	+	+	+	+
<b>Mode of termination</b>	MTP	MTP	MTP	MTP	MTP	MTP	MTP
<b>Fetal weight</b>	200	300	800	400	500	100	350

The incidence of congenital disorders is twice in preterm babies [14] With CNS abnormality this study is correlated with the study done by P. A. Boyd et al (2003) [15] and A.G. Tomatir et al (2009) [13] and urogenital abnormality this study is very well correlated with and sivasankara nayak (2015) [7]. and musculoskeletal system is correlated with study of A.G.Tomatir et al (2009) [13] and sivasankara nayak (2015) [7].

**Benefits of autopsy:** The direct benefits of autopsy to parents are not limited to refining the risk of recurrence. Even after autopsy, sometimes a definitive final diagnosis cannot be made and information given to parents may cover a range of possible diagnoses. In such cases the storage of fetal samples for possible future genetic analysis provides the hope of an accurate diagnosis (which may have ramifications for the wider family) at a much later date. In most cases in which the scan findings are confirmed parents can gain comfort that their baby had the prenatally suspected condition.

The finding of additional malformations, as well as in some cases changing the diagnosis, may be helpful in targeting tests in a subsequent pregnancy. A wider importance of autopsy is in its value for quality control for prenatal diagnosis, teaching, and research. The decline in autopsy rate and issues surrounding the retention of tissues and organs for diagnostic studies, teaching, and research has been the subject of much debate since the adverse publicity concerning autopsies and organ retention.

Parents should be provided with full information and not be coerced into accepting an autopsy examination. Parents need full information about the potential benefits of the examination, including details both about the procedures involved and about the benefits in providing information about risks of recurrence if they are to make a truly informed decision.

This discussion should be with an appropriately trained professional. Our study provides important information for parents. If a termination has been carried out because of anomalies detected by ultrasound scan, by declining an autopsy, parents will remain ignorant of information of recurrence risk.

## Conclusion

This study confirms the most number of perinatal deaths occurred in Low Birth Weight and preterm babies. Even though the prenatal ultrasonography reasonably predicts the malformations, fetal autopsy is essential to confirm the ultrasonogram diagnosis and also to look for additional malformations. Fetal and neonatal autopsy helps the parents by giving the information regarding recurrence risk of fetal anomaly, so that regular antenatal checkups with specific diagnostic test help to avoid congenital anomalies in subsequent newborn. Out of 7 fetal death 2 babies are born to primigravida and 5 babies to multigravida. All foetuses have congenital anomaly. Fisher P value  $<.05$ . 71.4% of fetus with weight less than 500 gm and 28.57% of fetus with weight range b/w 500-1000 gm have congenital anomalies and all congenital anomalies have birth weight less than 1000 gm. (Fisher Exact P value  $<.05$ ). All cases are 100% correlated with ultrasonogram.

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**Permission from IRB:** Yes

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