

Original Article

# Pattern and Prevalence of Congenital Malformation of fetus- Autopsy Based Study in BTGH (Basaweshwara Teaching and General Hospital) Mahadevappa Rampure Medical College(MRMCK). Kalaburgi. From Jan 2016- Jan 2017

Vandana Mudda<sup>1</sup>, Seemantkumar Saini<sup>2</sup>, Aditya Kumar Awati<sup>3</sup>

<sup>1</sup>Prof Dept. of FMT, <sup>2</sup>Post Graduate Dept. of Pathology, <sup>3</sup>Post graduate Dept. of Medicine, MRMCK. Kalaburgi

## Abstract

**Introduction:** Autopsy is an important aspect of clinical service, providing clinicians with critical feedback regarding diagnostic accuracy, therapeutic efficacy, and medical complication<sup>1</sup>

Among the Dravidian populations of the four southern states of India, Andhra Pradesh, Karnataka, Kerala, and Tamil Nadu, consanguineous marriages are<sup>5</sup> strongly favored which is one of the known cause of congenital malformation. Congenital malformation is emerging as an important component in the perinatal mortality and morbidity with considerable repercussion on the families' affected. Early diagnosis of life threatening congenital malformation can pave the way for surgical correction or palliation of these infants<sup>1</sup>.

**Material and Method :** We studied 217 cases of fetal autopsies from January 2016 to January 2017 duration of one-year retrospective study. Purpose of study is pathological and legal correlation in aspect of M: F ratio, age of termination of pregnancy, fetal anomalies & its pattern in Btgh Mrmck.Karnataka.

**Results:** Total fetal autopsies done are 217, out of which 51 are anomalous. Among them 22 are male and 28 are female babies and 1 sex is not determined {ambiguous}. In our study M: F ratio is 1:1.35. Most common cause of death found in autopsy examination is meconium aspiration in male fetus and placental insufficiency in female fetus. And mean age of gestation is 29 week and 30 weeks resp. Mean age of the mother is 29 years.

**Conclusion:** Fetal autopsies provide us an important information about pattern of anomalies, their incidence and cause of death in relation with sex and age of fetus, maternal age, along with socio-economic status. Legal implications regarding fetal autopsies is still a field of interest.

**Keywords** – Fetal autopsy, fetal anomalies, Legal aspects.

## Introduction

Fetal autopsy includes external, internal and histopathological examination of dead fetus along with placental examination<sup>2</sup>.

There are **two types** of fetal autopsy.

1. Medico-legal autopsy
2. Academic autopsy

**Medico legal Autopsy** –

Is conducted on requisition of police under section Crpc -174 to know the cause of death, age, sex and

---

**Corresponding author:**

**Dr. Vandana Mudda**

Prof Dept. of FMT. MRMCK. Kalaburgi-585105

viability of fetus for which police inquest & panchnama is required<sup>2</sup>

### **Academic Autopsy / Clinico-pathological Autopsy**

Is conducted on request of obstetrician, pediatrician, radiologist or family members of fetus to know the cause of congenital malformation and cause of repeated abortions, where in the detail pre and postnatal history of mother and consent of parent /relative with the collaboration of concerned department is necessary<sup>2</sup>.

In our study we conducted 217 fetal autopsies to rule out cause of death and associated anomalies which is helpful in future family planning for couples and improvement of management of anomalous babies

Etiological diagnosis in unexplained fetal deaths is possible with detailed evaluation of fetus. Fetal autopsy is confirmative in 28.6-89%, diagnostic in 10-38%; it provided additional information in 3.9-24% cases; it changed the predicted probability in 18% cases in addition, the data pertaining to demography, socio-economic status, and maternal health is helpful to pinpoint the factors behind the occurrence of fetal loss. At the Census of India conducted in 1981, the combined population of the four southern states exceeded 164 million, and consanguineous marriages are also common in the contiguous portion of Maharashtra.<sup>7</sup>

**Congenital anomalies were estimated to be the fifth largest cause of neonatal deaths in India after preterm births (34.7%), intrapartum complications (19.6%), pneumonia (16.3%) and neonatal sepsis (15%) Despite this ranking, in absolute numbers, congenital anomalies were estimated to contribute to 60699 neonatal deaths in India in 2013, which accounted for the highest global burden of neonatal mortality due to congenital anomalies India lacks national birth defects surveillance, indicating that there is no data on the magnitude of congenital anomalies in the country.<sup>7,8</sup> Thus, systematic data on the magnitude of congenital anomalies, the most prevalent types of congenital anomalies, their healthcare impact and their impact on neonatal health are required, especially as India has announced a program for the management of children born with selected birth defects like cleft palate lips etc.**

### **Material and Method**

We studied 217 cases. The present study of congenital anomalies in fetal and neonatal deaths was done at Mrmck. Study conducted over a time period of 1 year from January 2016 to January 2017. Consent for autopsy in requested compassionately, respectfully and was fully informed. Autopsy was within the scope of the autopsy permit and all the legal requirements are met before it is conducted. The autopsy protocol was including space for recording specific measurements and norms for particular gestational ages.

#### **Measurements:**

The crown heel length (CHL) and crown rump (CRL) length determined to the nearest 0.5 cm. Chest and abdominal circumferences were taken at the level of the nipple and umbilicus resp. Both limb measurement taken. The distances between the inner canthi and outer canthi, nasal height and width, philtrum height, mouth width and ear length are obtained and compared with published norms. All major organs weighed Photographs were taken

#### **Inclusion and exclusion criteria:**

The present study included dead fetus and neonates with gestational age 18 to 40 weeks of intra uterine life. All fetuses of gestational age <12 weeks and all neonates above 7 days of age were excluded from the study. Autopsy were 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. The Y shaped incision was taken. Umbilical vein examined for signs of inflammation, vernix, rupture (or) thrombus. The two umbilical arteries are examined and inspected in their entirety. The arteries and urachus examined for patency and arteries for hemorrhage (or) thrombosis. Single umbilical artery was an important anomaly noted. The autopsy protocol included the removal of thoracic, cervical, abdominal and pelvic organs en block and subsequently dissected into organ blocks<sup>3</sup>.

#### **Internal examination:**

All internal organs position and size and weight

were noted. The internal genitalia are inspected. As the testis will be undescended in younger fetuses, are removed with abdominal contents. Prior to opening of the pleural cavities the possibility of pneumothorax is ruled out. on entering the chest each cavity inspected for fluid. each lung was examined for developmental changes carefully. The integrity and tension of the pericardium are ascertained and the pericardial cavity is looked for the presence of free gas (or) fluid and fibrinous deposition. Heart examined in situ. All major veins and arteries examined. The diagnosis of premature closure of foramen ovale if any are noted. 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 part of heart, the interior of the left atrium<sup>3</sup>pulmonary venous orifices, mitral valve and left ventricle were inspected, followed by examination of the aortic valve and ascending aorta. Neck structures trachea and esophagus were examined. The scalp, fontanels, and cranial sutures were examined by palpation and changes documented. The fontanels, sutures, and glia were examined and changes documented. brain examined in situ. Then the brain was removed and examined and placed in fixative. Attention to the cranial base and dural sinuses was given<sup>3</sup>

### Dissecting the viscera

Examination begins with the most posterior structures and moved 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 are examined. The vagina and uterus are opened in the anterior midline and examined. The liver, gallbladder and structures of the porta hepatis, portal vein, hepatic artery and common bile duct are identified and dissected as indicated<sup>3</sup>.

The esophagus was opened in the posterior midline while intact with trachea. In this way a trachea-esophageal fistula can be identified and opened. Next the incision carried into the stomach. After major hilar structures of the lungs have been opened and inspected, attention was given to the lungs. Lobation and condition of the visceral pleura were presumably ascertained. In case of bladder outlet obstruction, the entire urethra is examined for posterior urethral valves (or) other abnormalities (i.e., anterior urethral valves, mega urethra). Placenta was

available in only few cases<sup>3</sup>.

### Histo-pathological 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 Hematoxylin and Eosin (H&E) stains. Special stains were done whenever necessary and studied. Autopsy findings were compared with ultrasound findings whenever available

## Results

**Table.-1. Percentage of fetal deaths (FD) early neonatal death (ND).**

Classification	No of cases	Percentage ( % )
Fetal Death (FD)	213	98
Neonatal death (ND)	04	02
Total	217	

**Table. – 2. Relation of maternal age (yrs.) with No of Fetal/Neonatal deaths.**

Maternal age (yrs.)	Fetal death	Neonatal death
No	No %	No %
19	3 1.38	
20 – 24	99 45.6	
25 – 29	81 37.3	3 1.38
30 – 34	23 10.5	1 0.5
35 – 39	07 3.22	
Total	213	

**Table – 3. External and Internal congenital anomalies.**

External congenital anomalies System affected	Type of anomaly	No	%
Neural and Spinal malformation.	Anencephaly	5	28
	Omphalocele	1	5.5
	Meningocele	5	28
	Hydrocephalous	2	11
	Acephalous	1	5.5
Lymphatic system	Hamartoma nape of neck	1	5.5
Skeletal system	Achondroplasia	2	11
Circulatory system	Single umbilical artery	1	5.5
Total		18	
Internal congenital anomalies.			
Respiratory system	Atelectasis of lungs	5	33
Genitourinary system	Polycystic kidney	2	13
Cardiac system	Tetralogy of Fallot	1	6.6
	Hypo-plastic heart	1	6.6
Others	Diaphragmatic hernia	6	40
Total		15	

**Table 4 - Fetal anomaly pattern according to sex distribution: In male fetus-22 cases (study of 217cases)**

S.No	Neural & spinal malformation. 8 cases	Cardiac malformn 3 cases	Pulmonary malformation 6 cases	Genitourinary Renal malform 1 case	Miscellaneous 4 cases
1	Anencephaly.	Myocarditis	Atelectasis	Poly cystic Kidney	Hydrops Fetalis.
2.	Anencephaly with Spinabifida	Tetralogy Of Fallot	Atelectasis		Chlongiomatous placenta
3.	Omphalocele.	Hypo-plastic Heart	Atelectasis		Edward syndrome (Trisomy – 18)
4.	Meningocele		Diaphragmatic Hernia		Edward syndrome (Trisomy – 18)
5.	Meningocele-Myelocele		Congenital Adenoid Cystic Malformn		
6.	Meningocele-Encephocele		Congenital Adenoid Cystic Malformn		
7.	Hydrocephalus				
8.	Hydrocephalus with spina bifida				

**Table 5 - Fetal anomaly pattern: In female fetus - 28cases (study of 217cases)**

S.No	Neural & spinal malformation. 8 cases	Cardiac malform 1 case	Pulmonary malformation 7 cases	Pulmonary malformation 6 cases	Miscellaneous 6 cases
1	Anencephaly – 3cases	Ebstein Anomaly	Partial Atelectasis – 2cases	Urethral Stenosis	Sacrococcygeal teratoma
2	Anencephaly with Spinabifida- 2case		Diaphragmatic Hernia – 2cases	Extrophy Bladder	Hamartoma of Nape of neck
3	Anencephaly with spinal deformity		Diaphragmatic Hernia with CCAM Type II– 2cases	Winters-syndrom Renal Dysplasia	Achondrodysplasia – 2cases
4	Acephalous		Congenital Adenoid Cystic Malformation TYPE III	Bilateral Renal Cystic Diseases	Klipilfel-Syndrom 2 cases.
5	Meningocele-occipital			Mermaid – Renal Agenesis	
6				Renal – Hepatic Splenic Dysplasia	

\* **THANATROPIC DYSPLASIA 1 case (Sex not Determined), M: F- 1:1.3, Total anomalous–51 cases.**

**Table 6 -Internal congenital anomalies – Relation between maternal age, fetal weight and fetal anomaly.**

Total anomaly	Mother Age ( Yr.)	Fetal anomaly	Fetal weight (Mean wt.)	
			Total	
			47cases	04 cases
51 cases	25-29	20	1000 gms	
	30-34	24	800 gms	2.8 kgs
	35-39	07	750 gms	

## Discussion

Fetal autopsy significantly contributes to the diagnosis of intrauterine fetal death and congenital anomalies as a major cause of perinatal death. Congenital malformations in fetal and neonatal deaths vary in different studies. The study of malformations greatly helpful in genetic counseling and prenatal diagnosis in successive pregnancies<sup>1</sup>.

In the present study 217 fetal and neonatal autopsies were carried out among 213fetal and 04 neonatal deaths that occurred in Btgh , during the period from JAN 2016 to JAN 2017. Prevalence of Congenital malformations account for 23.5% of fetal and neonatal deaths. This incidence matches with the study by Rabah M. Shawky

, Nermine S. Elsayed

**MATERNAL FACTORS:** In present study, the incidence of congenital malformations was higher in mother's age group of 25 to 39 years. In 25 to 29 years out of 217 fetal and neonatal deaths 20 (9.2%) cases got anomalies. In 30 to 34 years of maternal age group, 24 (11.05%) cases got anomalies. In 35-39 years of maternal age group, 07 (3.2%)cases got anomalies Many authors have shown higher incidence of malformations in the babies born to maternal age betn 20 to 35 years. The observations in the present study is that 90% of the cases belong to multigravida and 10% cases belong to primigravida. In our study the incidence of congenital anomalies are increased with the increase in maternal age

**FETAL FACTORS:** -In present study, the incidence of congenital malformations were higher among the low birth weight infants (<1500 gm) in comparison to the normal weight accounting for 04 cases. The association of low birth weight and malformations has been well documented. Many studies have documented male predominance amongst congenital malformed babies. However, in the present study we observe 22 male babies and 28 female babies with congenital malformation. In a five year study on major congenital anomalies in Turkey by Tomtair et al.,<sup>14</sup> there were 183 cases (2.9/1000) of single (or) multiple congenital Anomalies among 63,159 live births. The most common anomalies were related to the nervous system (31.1%), cleft palate and lip (18.6%), musculoskeletal system disorder (14.2%) and chromosomal anomalies (13.1%). Both genders were found to have greater anomalies related to the nervous system (34.9% of girls and 28.3% of boys) while amongst.

In present study CNS malformation is the most common. (17cases 8%) in 217 cases. that follows the order Meningocele > Anencephaly > Hydrocephalus. Second most common malformation is pulmonary malformation (13cases 6%) in 217 cases, most common is Diaphragmatic hernia than Atelectasis of Lung. Urogenital malformation (7cases 3.2%) in 217 cases. – With female predominance.

#### **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<sup>5</sup>.

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. It is important that those advising them at such a sensitive time do not take what may be the superficially kinder route of avoiding detailed discussion about the autopsy. 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<sup>5</sup>.

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**

The study of dead is to save the livings. Congenital malformations have become important cause of fetal and neonatal mortality in developed countries and would very soon be increasingly important determinants of fetal and neonatal mortality in developing countries like India and of various states- like Karnataka where consensual marriage is common and is known cause of congenital malformation This study was undertaken with the purpose of finding out cause of death during the perinatal & neonatal period at Btgh, mrmc kalaburagi, Karnataka, to see pattern and prevalence of congenital anomalies and implication of legal aspects of fetal autopsy.

**Conflict of Interest-** No

**Source of Funding-** Personal

**Ethical Clearance:** Not necessary

#### **References**

1. Mariana costache, anca mihaela lazarioiu, andreea contolenco. Clinical or Postmortem? The Importance of the Autopsy; a Retrospective Study. A Journal of Clinical Medicine. 2014; 9(3):261-5.
2. Cristoforo pomara, StevenB karch, Vittorio Fineschi. Forensic Autopsy. A Hand Book and Atlas. 2010; 1:200-4.

3. Ludwig J. Principles of autopsy technique immediate and restricted autopsy and other special procedure. Ludwig J Handbook of Autopsy Practice. 2002; 3:3-6.
4. Park K. Congenital malformations. Park's Textbook of Preventive and Social Medicine. Banarasidas Bhanot Publications Jabalpur.2005;18:379-80.
5. Siva Sankara Naik V et al. Int J Res Med Sci. 2015; 3(5):1114-21.
6. Grover N. Congenital malformations in Shimla. Indian J Pediatrics. 2000;67(4):24-5.
7. Seshadri S, Guruswamy T, Jagadeesh S, Suresh I. Methodological issues in setting up a surveillance system for birth defects in India. Natl Med J India. 2005; 18:259-62.
8. Radha Rama Devi A, Appaji Rao N, Bittles AH. Inbreeding in the state of Karnataka. South India. 1982; 32:8-10.
9. Kumar S Pai, RA Swami Nathan, MS. Consanguineous marriages and the genetic load due to Lethal genes in Kerala. AnnHumn Gentet 1967; 31:141-5.
10. Rave AK. Nature, amount and extent of consanguinity among two South Indian castes. J Hered 1979;70:281-6
11. Centerwall WR, Savarinthan G, Mohan LR, Booshanan V, Zachariah M. Inbreeding patterns in South India. Soc Biol 1969; 16:81-91.