Original Article

Indian Journal of Forensic Medicine and Pathology
Volume 12 Number 1, January - March 2019
DOI: http://dx.doi.org/10.21088/ijfmp.0974.3383.12119.6

Pattern and Prevalence of Congenital Malformation of Fetus: Autopsy Based Study

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Abstract

Introduction: Autopsy is an important aspect of clinical service, providing clinicians with critical feedback regarding diagnostic accuracy, therapeutic efficacy, and medical complication [1]. Rajasthan has reportedly the second Indian state having highest number of child marriages in the country with more than 60 percent of girls getting married before 18 [1]. The practice of child marriage is common in Rajasthan. Along with cultural incest marriage leads to one most important causes of fatal anomalies. Early diagnosis of life threatening congenital malformation can pave the way for surgical correction or palliation of these infants [1].

Material and Method: We studied 217 cases of fetal autopsies from January 2017 to October 2018 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 Pacific Institute of Medical Sciences (PIMS) and M.R. Medical College, Gulbarga, Karnatka.

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 respectively. 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-economical status. Legal implications regarding fetal autopsies is still a field of interest.

Keywords: Fetal Autopsy: Fetal Anomalies: Legal Aspects.

How to cite this article:
Seemant Kumar Saini, Matariswa Samanta, DR Mathur, et al. Pattern and Prevalence of Congenital Malformation of Fetus: Autopsy Based Study. Indian J Forensic Med Pathol. 2019;12(1): 29-36.

Introduction

Fetal autopsy includes external, internal and histopathological examination of dead fetus along with placental examination [2].

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Received on 27.11.2018, **Accepted on** 28.12.2018

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 viability of fetus for which police inquest& panchnama is required [2].

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 if any and cause of repeated abortions, where in the detail pre and post natal history of mother and consent of parents /relatives with the collaboration of concerned department is necessary [2]. Placental examination is done to know the cause of death to certain extent and placental pathology explains the cause of prematurity.

In our study we performed 217 cases of fetal autopsy 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.

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 [7,8]. 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 me for the management of children born with selected birth defects like cleft palate lips etc. [9,10].

Materials and Method

We studied 217 cases The present study of congenital anomalies in fetal and neonatal deaths was done at PIMS. Study conducted over a time period of 8 month from January 2017 to october 2018. 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 dissecting instruments required for fetal and neonatal (perinatal) autopsy are small scissors and forceps and scalpels. The autopsy protocol was including space for recording specific measurements and norms for particular gestational ages.

Measurements

The crowns heal 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 respectively. Both limb measurements has been taken. The metric documentation of changes in the face is often a valuable component of the autopsy. So 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. Weights: Scales accurated to 0.1 gm.in perinatal specimens. All major organs 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 which provide indisputable evidence of findings and study of dysmorphic face images, for important diagnostic information [3].

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 was performed by standard technique adopted by Edith L. Potter.

Procedure

1-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 taken which extend from the anterior aspect of each shoulder to the xiphoid process. 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 the 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 [3].

2- Internal examination

All internal organs position and size and weight were examined. 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 pneumo thorax 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, while anatomic relationship with structures were intact, inspected externally and internally, in a systematic fashion that follows the flow of blood. All major veins and arteries were 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 [3] 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 en bloc by the rokitansky methods of evisceration. Neck structures trachea and esophagus were examined. The scalp, fontanels, and cranial sutures were examined by palpation and any changes were documented. The fontanels, sutures, and glia were examined and any changes were documented. brain has been exposed and examined in situ. Then the brain was removed and examined on all sides and placed in fixative. Attention to the cranial base and dural sinuses was given [3].

3- 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 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 [3].

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 turned 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 [3].

4-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 Haemato-xylin and Eosin (H&E) stains. Special stains were done whenever necessary and studied. Autopsy findings were compared with ultrasound findings whenever available.

Results

In present study out of 217 patients 213 (98%) were fetal death and 4 (2%) cases of neonatal death were included in present study. Among 217 cases 51 found to anomalous. Relation between maternal age and neonatal death mean age of mother was 29 years. 99 (45.6%) fetal death seen in maternal age group was 20-24 years. While in neonatal death maternal age most commonly involved was 25-29 years.

External anomalies most commonly seen in present study was Neural and spinal malformations that included 77% of external anomalies. While Lymphatic system & Circulatory system involvement was only 5.5% in present study.

In Internal congenital anomalies diaphragmatic hernia (40%) was most common anomaly seen followed by atelectasis of lung i.e. 33% in present study. Cardiac and genitourinary anomalies involvement was 13% respectively.

Relationship between anomalous fetus and weight of fetus reveals that 47 anomalous fetus had weight of 1000 gm, 800 gm & 750 gm respectively while 4 fetus had normal weight range 2.5 to 3.5 kg. And 30-34 year of maternal age involved maximum number of anomalous fetus which was 47% seen in present study.

Anomalous pattern in 22 male fetus involved Neural and spinal Malformation (8 cases) > Pulmonary malformation (6 cases) > cardiac malformations (3 cases) > genitourinary & Renal malformation (1 case) and in miscellaneous 4 cases

2 cases diagnosed as Edwards syndrome. while anomalous pattern in female fetus included no of cases were 28, female fetus it s bit higher incidence as compare to male fetus anomalies ratio of anomalies in study was 1:1.3, Neural and spinal Malformation (8 cases) > Pulmonary malformation (7 cases) > Genitourinary & Renal malformation (6 cases) > cardiac malformations (1 case). In miscellaneous category total number of cases were 6 cases among them 2 cases diagnosed as Klipilfeil Syndrome & 2 cases diagnosed as Achondrodysplasia in present study. Thanatropic dysplasia seen in 1 case (sex was not determined). As we mentioned consensual marriage incidence in our study is 83%, 180 cases had history of consensual marriage a great significance in formation of anomalous fetus pattern and preventable cause by spreading awareness among population subgroups. There is no toxicological study done in present study due to lack of any relevant significance because cause of death known in all cases as malformation.

Table 1: Percentage of fetal deaths (FD) and early neonatal death (END).

Classification	No. of cases	Percentage (%)
Fetal death (FD)	213	98
Neonatal death (ND)	04	2
Total		

Table 2: Relation of maternal age (yrs) with no of fetal/neonatal deaths

Maternal age (years)	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
s35-39	07	3.22		
Total	213		4	

Table 3: External congenital anomalies

System affected	Type of anomaly	No	%
Neural &spinal malformation	Anencephaly	5	28
	Omphalocele	1	5.5
	Meningocele	5	28
	Hydrocephalus	2	11
	acephalus	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	

Table 4: Internal congenital anomaly

	Type of anomaly	No.	%
Respiratory system	Atelectasia of lungs	5	33
Genitourinary system	Polycystic kidneys	2	13
Cardiac system	Tetrology of Fallot	1	6.6
	Hypoplastic heart	1	6.6
others	Diaphragmatic hernia	6	40
Total		15	

Table 5: Relation between Maternal age , Fetal weight and fetal anomaly

Total anomaly	Mother Age (Yr)	Fetal anomaly	Fetal weight (Mean wt)	
51 cases			Total no case-47	Total no of case – 04 case
	25-29	20	1000 gm	
	30-34	24	800gm	2.8 kg
	35-39	07	750gm	

* 04 Cases Having Normal Weight Range 2.5 -3.0

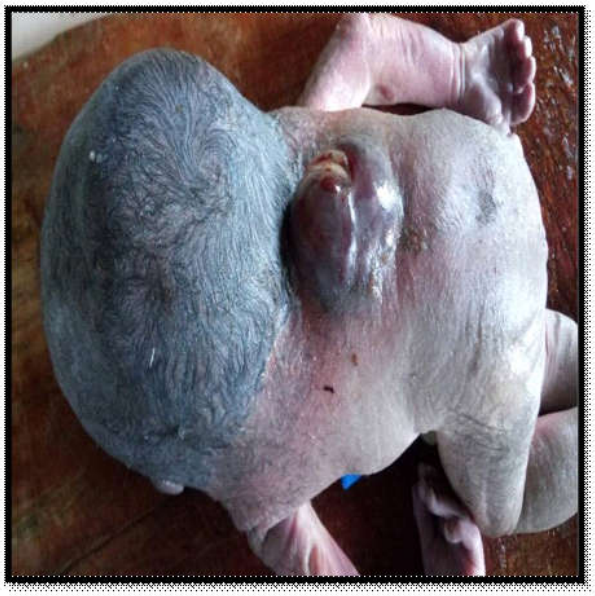
Table 6: Fetal Anomaly Pattern According to Sex Distribution Anomaly Pattern in male fetus-22 Cases (Study of 217 Cases)

Sno.	Neural & Spinal Malformation 8 Cases	Cardiac Malformation 3 Cases	Pulmonary Malformation 6 Cases	Genitourinary / Renal Malformation 1 Case	Miscellenous 4 Cases
1	Anencephaly -1	Myocarditis-1	Atelectasis – 3 cases	Poly cystic Kidney -1	Hydrops Fetalis-1
2	Anencephaly with Spinabifida-1	Tetrology of Fallot -1	Diaphragmatic Hernia – 2cases		Chlongiomatous placenta-1
3	Omphalocele-1	Hypoplastic Heart -1	Congenital Adenoid Cystic Malformation-1		Edward syndrome (Trisomy – 18) – 2 cases
4	Meningocele -1				
5	Meningocele-Myelocele-1	-	-	-	-
6	Meningocele Encephocele-1	-	-	-	-
7	Hydrocephalus-1	-	-	-	-
8	Hydrocephalus with Spinabifida-1	-	-	-	-

Table 7: Anomaly pattern in female fetus study of (total 217 cases) – 28 cases

S.No.	Neural & Spinal Malformation -8 Cases	Cardiac Malformation-1 Case	Pulmonary Malformation-7 Cases	Genitourinary / Renal Malformation-6 Cases	Miscellenou-6 Cases
1	Anencephaly – 3cases	Ebstein Anomaly	Partial Atelectasis – 2cases	Uretheral 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 Syndrome – Renal Dysplasia	Achondrodysplasia – 2cases
4	Acephalous		Congenital Adenoid Cystic Malformation TYPE III	Bilateral Renal Cystic Diseases	Klipilfeil Syndrome – 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



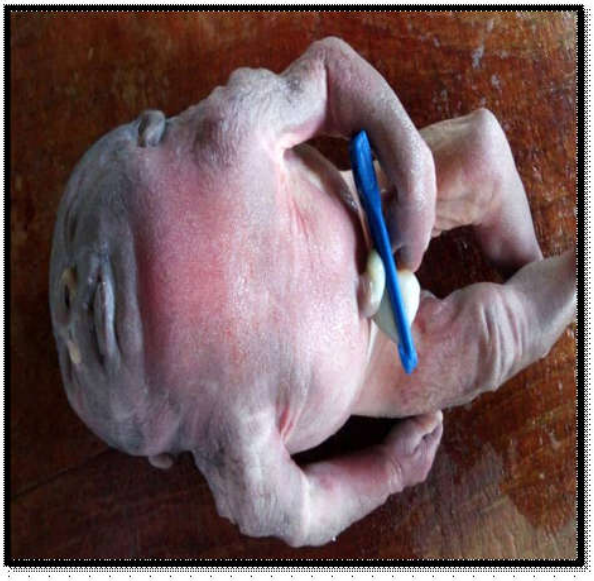


Fig 1: Gross – fetus with short neck (web neck) and meningocele

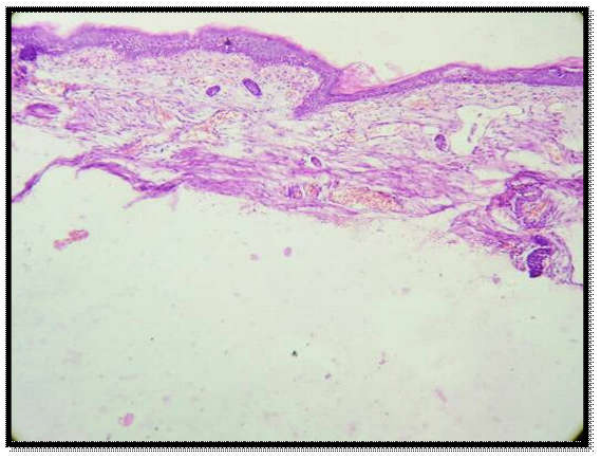


Fig. 2: H & E (400x) section reveals meninigocele-showing kertinized stratified squamous epithelum with neural tissue

Fig. 3. Gross–lung showing multiple cystic lesion

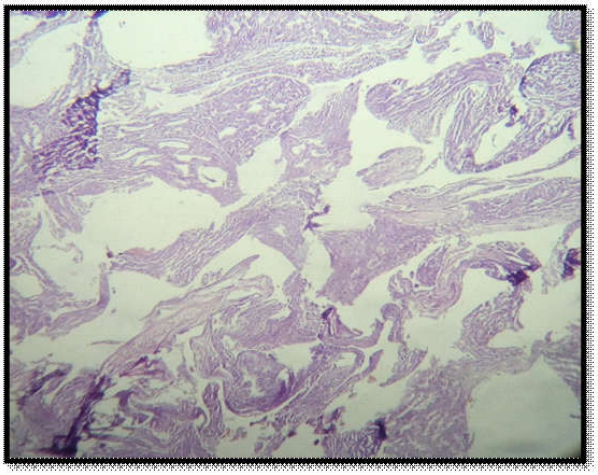


Fig.4: H & E (400x) section showing congnital cystoid malformation type-1

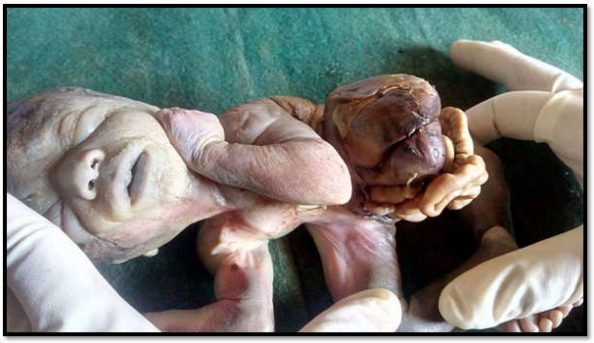


Fig. 5: Gross - fetus showing polycystic kidney &omphelocele

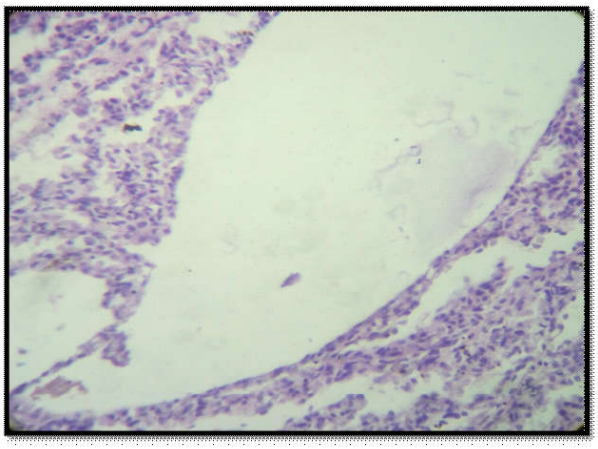


Fig. 6: H&e (400 x) section showing multiple cystic lesion - polycystic kidney

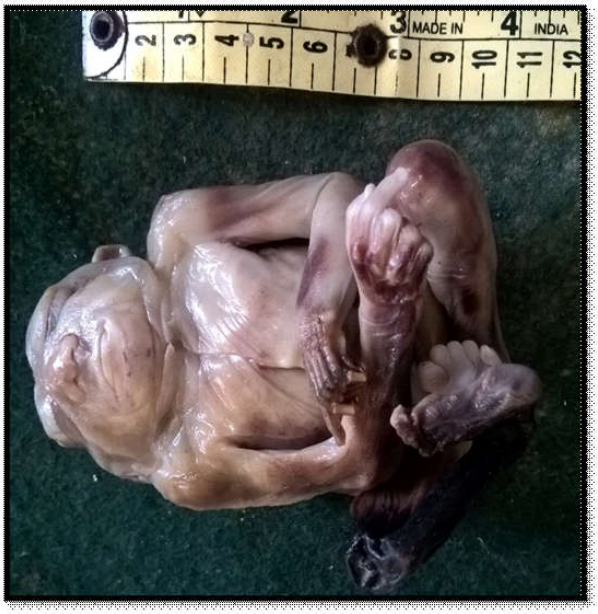


Fig. 7: Gross picture fetus showing microcephaly

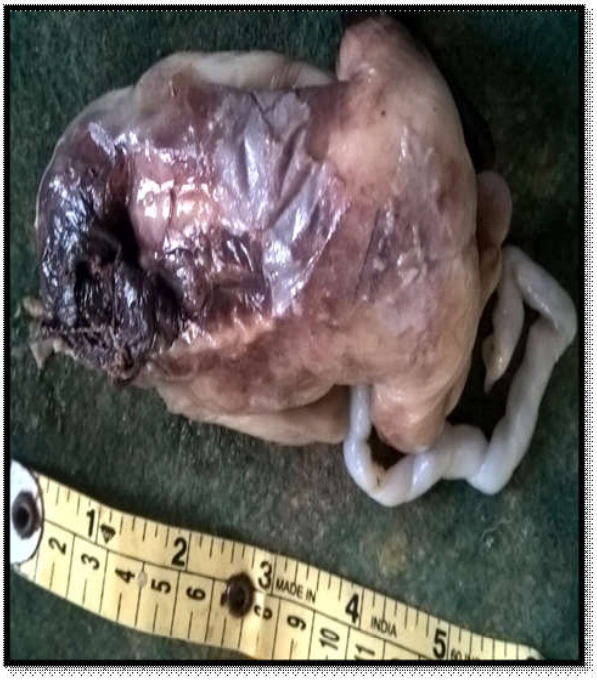


Fig. 8: gross picture fetus showing meningocele

Discussion

Fetal autopsy significantly contributes to the diagnosis of intrauterine fetal death and congenital anomalies are 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 pregnancies1.

In the present study 217 fetal and neonatal autopsies were carried out among 213 fetal and 04 neonatal deaths that occurred in PIMS, during the period from Jan 2018 to Oct 2018. 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 were 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 between 20 to 35 years. The observations in the present study is that 90% of the cases belong to multi gravid and 10% cases belong to primi gravida. In our study the incidence of congenital anomalies were 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., 14 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. Among them most common follows the order Meningocele > Anencephaly > Hydrocephalus. Second most common malformation is pulmonary malformation (13 cases 6%) in 217 cases ,most common is Diaphragmatic Hernia than Atelectasis of Lung. Urogenital malformation (7 cases 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 [5].

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 [5].

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 Rajasthan, Karnataka & Andhra Pradesh where consensual marriage is common which 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 PIMS Udaipur, to see pattern and prevalence of congenital anomalies and implication of legal aspects of fetal autopsy.

Confliction of interest: No

Ethical clearance: Not necessary.

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