Original Article

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Pattern of Congenital Anomalies: A Hospital Based Fetal Autopsy Study

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Abstract

Congenital malformations are most common cause of perinatal death. Antenatal imaging studies can easily detect the problem, and can give an idea of diagnosis. However autopsy still plays a pivotal role in confirming the diagnosis and identifying actual cause of fetal loss. *Materials And Methods:* This is a descriptive, cross-sectional study on 101 fetuses, over a period of 2 years from October 2014 to September 2016 referred to the Department of Pathology, Dr PSIMS & RF; which were an outcome of spontaneous abortions, intrauterine deaths, still births and of therapeutic abortions due to anomalies. This study is to determine overall incidence and distribution of various congenital anomalies and data was analyzed statistically. *Results:* Total 101 consecutive perinatal autopsy were performed, 31 (30.6%) fetuses showing congenital anomalies with female (16.8%) predominance and M:F ratio 1:1.2. Common congenital anomalies were observed in this study included central nervous system defects - anencephaly and meningomyelocele being common. Other systemic anomalies are genitourinary system, gastrointestinal system, respiratory and musculoskeletol system were seen. Multiple syndromes such as Prune-belle, Meckel- gruber were observed in this study.

Keywords: Perinatal Autopsy; Congenital Anomalies.

Introduction

Congenital Malformation can be a physical, metabolic or anatomic defect which may be evident before birth, at birth, or detected during the first year of life. The malformations can present itself in a single organ, single system, or may involve multiple organs of the body [1]. Congenital malformations remain a common cause of fetal deaths and accounts for 25-30% of fetal deaths in developed countries and 10-15% of fetal deaths in developing countries like India [2].

The malformations can be detected prenatally by ultrasound, maternal serum analysis etc.; and by autopsy after the fetal death. Fetal autopsies can provide a diagnostic clue to determine the cause of death. Various studies on fetal autopsy was found to be confirmative in 28.6-89%, diagnostic in 10-38%; provided additional information in 3.9-24% cases; and it had changed the predicted probability in 18% cases [3].

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Aims and Objectives

To represent an analysis of autopsy findings including congenital anomalies of fetuses and to study clinical and pathological findings (Gross & microscopic) in fetal death referred to department of pathology, Dr. PSIMS & RF.

Materials and Methods

This was a descriptive, cross-sectional study on 101 fetuses, over a period of 2 years from October 2014 to September 2016 referred to the Department of Pathology, Dr PSIMS & RF; which were an outcome of spontaneous abortions, intrauterine deaths, still births and of therapeutic abortions

due to anomalies. Maternal history, age and sex of the fetus were also recorded. Verbal and written consent was taken from the mother.

Autopsy was performed by standard technique adopted by Edith L. Potter. Each fetus was examined according to a predesigned protocol which included photograph, morphometric parameters, external examination, and internal examination by giving a straight line incision starting from chin to pubic symphysis passing to the left of the umbilicus. Skin and subcutaneous tissue was retracted. Cervical, thoracic, abdominal, pelvic cavities were opened and any deviation from the normal anatomy noted. The autopsy protocol included the removal of thoracic, cervical, abdominal and pelvic organs en block and subsequently dissected into organ blocks. Tissues from the organ blocks were sent for histopathological examination. The placenta, fetal membranes and umbilical cord were also studied. All the available information was correlated to ascertain the cause of death.

The types of birth defects were classified according to International statistical classification of diseases and related health problems, 10th revision (ICD-10). The study was approved by the Institutional Ethics Committee (Dr PSIMS &RF).

Results

Out of total 101 fetal autopsies, 51 (50.49%) specimens were from intrauterine deaths, 38 (37.62%) were from induced abortions followed by 12 (11.88%) specimens from neonatal deaths.

A total 101 fetal autopsies, 44 were males and 57 were female fetuses; out of which the incidence of congenital anomalies in males and females was 13.8% and 16.8% respectively.

When the maternal age was taken in to account, most of the fetal deaths were seen in mothers in the age group of 21-30 years corresponding to 72.27% of cases. Out of these cases 23.7% presented with anomalies.The incidence of fetal deaths was highest in primigravida which accounted for 76.2% of cases.

Out of 101 foetal autopsies, Congenital anomalies were seen in 31 cases which accounts for 30.6% of fetal deaths. Most of the cases were that of the central nervous system, (11cases, and 35.48%), of which anencephaly and myelomeningocele were the predominant.

The second most common congenital malformations were seen in genitourinary system (16.12%), followed by gastrointestinal system (12.9%), cardiovascular system (6.45%), respiratory and musculoskeletal system (3.22%). In the miscellaneous group (22.58%) 7 cases were encountered which included 2 cases of single umbilical artery, 2 cases of cystic hygroma, 1 case of diaphragmatic hernia, and 2 cases with multiple anomalies, leading to probable syndromes like Prune Belle Syndrome and Meckel Gruber Syndrome.

Table 1: Incidence of fetal & neonatal deaths

	No. of cases	Percentage%
Intrauterine deaths	51	50.49
Induced abortions	38	37.62
Neonatal deaths	12	11.88
Total	101	100

Table 2: Congenital anomalies in relation to sex

Anomaly	Sex		Total
	Male	Female	
Absent	30	40	70
Present	14 (13.8%)	17 (16.8%)	31
Total cases	44	57	101

Table 3: Congenital anomalies encountered in autopsies as per maternal age

Maternal age in years	Anomaly		Total
	Absent	Present	
<20	20	4	24
21-30	49	24 (23.76%)	73 (72.27%)
>30	1	3	4
Total	70	31	101 (100%)

Table 4: Congenital anomalies encountered in autopsies as per gravid

Gravida	Anomaly-Absent	Anomaly-Present	Total
Primi	52	25	77 (76.2%)
Second	8	4	12
Third	6	1	7
Fourth	3	0	3
Fifth	1	0	2
Total	70	31	101 (100%)

Table 5: System wise distribution of congenital anomalies

	Type of anomaly	No : 31	Percentage 100 %
1	CNS	11	35.48
2	GUT	5	16.12
3	GIT	4	12.9
4	RS	1	3.22
5	CVS	2	6.45
6	MSK	1	3.22
7	Miscellaneous with Multiple anomalies	7	22.58

Table 6: List of anomalies

Anomalies of central nervous system	
Type	**No. of cases**
Anencephaly	3
Myelomeningocele	3
Hydrocephaly	1
Choroid plexus cyst	1
Meningocele	1
Lissencephaly	1
Acrania with amniotic bands	1
Anomalies of Gastro intestinal system	
Type	**No of cases**
Gastroschisis	2
Omphalocele	1
Cleft lip and cleft palate	1
Anomalies of the Genitourinary system	
Bilateral Multicystic Renal dysplasia	3
Bilateral Renal Agenesis	1
Single kidney	1
Anomalies of Cardiovascular system	
Cardiac Hypoplasia	1
Cardiomegaly	1
Anomalies of Musculoskeletal system	
Arthrogryposis multiplex congenita	1
CTEV	2
Anomalies of Respiratory system	
High airway obstruction syndrome	1
Miscellaneous with multiple anomalies	
Diaphragmatic hernia	1
Cystic hygroma	1
Cases with Multiple anomalies	2
Potter sequence	1
Probable Syndromes considered	
Meckel Gruber Syndrome	1
Prune Belly Syndrome	1

Table 7: Number of Anomalies Comparison with other studies

System involved	Sunethri et al [5]	Datta et al [9]	Present study
Central nervous	12	5	11
Genitourinary	7	3	5
Gastrointestinal	8	8	4
Cardiovascular	1	2	2
Respiratory	0	0	1
Musculoskeletal	0	3	1
Miscellaneous	0	24	5
Syndromes	0	3	2
Total	28	48	31

Fig. 1: Anencephaly

Fig. 2: Potters Sequence(external rotation of both lower limbs, low set ears, pre auricular tags, depressed nasal bridge, with multi cystic renal dysplasia in box)

Fig. 3: multiple anomalies such as absence of two digits on right foot, imperforate anus, absent spleen, single enlarged multicystic dysplastic kidney and single umbilical artery

Discussion

Over a ten year period, there has been a decline in the number of fetal autopsies. Fetal autopsy, however was found to contribute significantly to the diagnosis of Intrauterine fetal death and to detect congenital anomalies [4]. Hence the present study was an attempt to study various parameters responsible for still birth, intrauterine death or therapeutic cause for termination and significance of autopsy in making final diagnosis.

In the present study on 101 fetal autopsies, 31 cases (30.6%) had showed congenital malformations. This incidence was slightly higher in comparision with the study by Sunethri et al [5] where it was 27%, and was less with the incidence observed by Harsha Mohan et al.[6] (38.7%). In the present study CNS malformations were most common Majority were due to defective closure of neural tube between the 23rd and 26th day of gestation resulting in anencephaly or meningomyelocele. Out of 3 cases of anencephaly, one case was associated with cleft lip and cleft palate. Our study is in accord with the study of Tomatir AG et al.[7]. (31.1%), and Swain et al.[8] (39.5%). Out of 3 cases of myelomeningocele, one case was associated with bilateral CTEV and had a history of Arnold –Chiari malformation.

The second most common congenital anomalies were encountered in genitourinary system. The present study coincides with the observations of Sankar et al.[2] (17.2%). Multicystic renal dysplasia was the most common presentation of which one case was presented with cystic hygroma. Gastrointestinal system anomalies (12.9%), in which gastroschisis was the most common, of which one case was associated with cystic hygroma and absent right lower limb.

The observation of the present study was also compared with studies of Sunethri et al. [5] and Datta et al. [9] and was found to accord more with the observations of Sunethri et al. [5].

A total of five cases with multiple anomalies were encountered and we discuss them. The first case was a 17 weeks female fetus born to 24 year old women with history of severe oligohydramnios had external rotation of lower limbs, low set ears, pre auricular tags, depressed nasal bridge and multi cystic renal dysplasia resulting in Potters Sequence.

The second case was a 13 week dead fetus with distended abdomen with deficient anterior abdominal wall muscles and imperforate anus. On opening the abdomen, bladder was markedly distended and measured 4 cm in diameter and was filled with clear fluid. Both the right and left kidneys showed total replacement of the renal parenchyma by the dilated pelvicalyceal system along with bilateral cystic renal dysplasia. The findings were suggestive of Prune belly syndrome.

The third case was a 19 weeks female fetus with multiple anomalies such as post axial polydactyly, single umbilical artery, absent anal opening, single fissure in right lung, incomplete left lung fissure, absent left kidney, and single ectopic kidney with cystic renal dysplastic changes. Possible syndrome considered was Meckel Gruber Syndrome, though we did not see classical meningocele association and hence advised cytogenetic analysis for further confirmation.

Fourth case was a 12 weeks male fetus born to 22 year old women with anterior abdominal wall defect, absent right limb, with cystic hygroma. Fifth case was a 19 weeks terminated male fetus born to 29 year old women with history of severe oligohydramnios had showed multiple anomalies such as absence of two digits on right foot, imperforate anus, absent spleen, single enlarged kidney with multicystic renal dysplasia and single umbilical artery.

Conclusion

Foetal death is common clinical problem and the family seeks and deserves answers regarding the cause of death. The findings shall helps in parental counseling and future pregnancy planning.

Now a days, merely decline in autopsy rates, continous presentation and publication of the fetal autopsy findings shall rejuvinate the dying science and create awareness to encourage foetal autopsies and genetic studies by Gynaecologists, Radiologists, Paediaticians, and Pathologists as a team work.

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