A STUDY OF INTERNAL MALFORMATIONS IN MEDICALLY TERMINATED AND IN STILLBORN ANOMALOUS FOETUSES IN A TERTIARY CARE HOSPITAL

Dr. Gnanavel A[1*], Saravana Kumar S[2],

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Abstract

Introduction:

Congenital malformations have become important cause of foetal and neonatal (perinatal) mortality in developed and developing countries The foetal autopsy plays the vital role in the confirmation and identification of associated congenital anomalies and also for the counselling of the parents, to prevent the further foetal congenital anomalies in subsequent pregnancies. The aim of our study is to do a complete external examination and dissection of terminated and stillborn anomalous foetuses to identify the internal anomalies associated with external malformation.

Methods:

This study was done at Meenakshi Medical College Hospital & Research Institute, Kancheepuram over a time period of 1 year. The present study included terminated and stillborn foetus obtained from mothers of bad obstetric history .The foetal autopsy was performed by standard technique. External and internal findings were observed.

Results: Out of 12 autopsies (7 male and 5 female) were performed, most common congenital anamolies in our study were anencephaly and meningomyelocoele, 6 foetus shows internal abnormalities. Malformations of urinary system were most commonly seen followed by abnormalities of liver, spleen and adrenal gland.

Conclusion: Majority of the foetal and perinatal death occur due to major congenital malformations and its study highlight the importance of autopsy and greatly helpful in genetic counselling and prenatal diagnosis in successive pregnancies.

Keywords: Foetus, Autopsy, Congenital anomalies

[1*]Associate professor, Department of Anatomy, Meenakshi Medical College Hospital & Research Institute, Meenakshi Academy of Higher Education and Research MAHER (Deemed to be University)

[2]Assistant professor, Department of Anatomy, Meenakshi Medical College Hospital & Research Institute, Meenakshi Academy of Higher Education and Research MAHER (Deemed to be University)

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1. Introduction

Congenital anomalies are considered as the major health problems that are responsible for the morbidity and mortality in newborns. It affects 2.5% of total live births in Indian population. Congenital anomalies account for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India.[1] According to WHO Congenital anomalies are defined as structural or functional anomalies, including metabolic disorders which are present at the time of birth.[2]

Around 40%- 60% of congenital anomalies are of unknown etiology.[3,4] anomalies with multifactorial cause 20-25%.10-13% of environmental and 12-25 % of genetic causes. 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. Antenatal sonography developed in recent years; however it continues to lag behind a complete foetal autopsy in accurately diagnosing the cause of foetal death. Only few studies have comparatively examined prenatal ultrasound findings and postnatal autopsy results [5]. The autopsy serves a vital and varied purpose in modern medicine, not the least of which is its role as a tool for diagnosis quality control and verification, which has a positive effect on clinical practice in the long run. [6].

Aims and objectives

To study the various internal malformation in terminated and stillborn anomalous foetuses.

To take appropriate preventive aspects of congenital anomalies.

2. Materials & Methods

The present study was done atMeenakshi Medical College Hospital & Research Institute, Kancheepuram. The study was conducted over a time period of 1year . The dissecting instruments required for foetal and neonatal (perinatal) autopsy are small scissors and forceps and scalpels. The standard autopsy protocol was followed. The present study included all stillborn and terminated foetus, autopsy was performed by standard technique adopted by Edith L. Potter. A thorough external examination was conducted to look for wounds and macerations, skin lesions and all major and minor developmental anomalies. An incision in the shape of a Y was made that ran from the front 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 block and subsequently dissected into organ blocks [5]. Examination begins with the most posterior structures and moves anteriorly layer by layer. 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 [7].

Inclusion criteria:

1. All congenital anomalous foetuses detected by USG and terminated.
2. Stillborn anomalous fetuses

Exclusion criteria:

1. Normal stillborn foetuses
2. Those who do not give consent for this study

3. Results

A total of 12 autopsies were performed, out of 7 cases were male and 5 cases were female and regarding the parity of the mother 8 were primi and 4 were multigravida. On external examination all the foetus showed congenital malformations. Most common congenital anomalies in our study are anencephaly and meningomyolocoele. On internal examination 6 foetus had internal congenital anomalies. Malformations of urinary system were most commonly seen followed by abnormalities of liver, spleen and adrenal gland.

TABLE 1: Types of anomalous foetus

External malformation	**Sex of foetus**		**Total**
	Male	**Female**	
Skeletal Dysplasia	2	1	3
Anencephaly	1	2	3

Meningomyelocoele	2	1	3
Meningocoele	1	1	2
Occipital encephalocoele	1	0	1
Total	7	5	12

In 3 anencephaly foetus the following congenital anomalies were adrenal gland agenesis, dysplastic kidney and huge hepatomegaly, in 3 meningomyelocoelefoetus show lobulated kidney, adrenal gland agenesis and asplenia, in 3 cases of skeletal dysplasia show dysplastic kidney and hypertrophy of thymus. Dysplastic and lobulated kidney were seen in occipital encephalocoele and no internal anomalies seen in meningocoele.

Figure 1: skeletal Dysplasia

Figure 2: Anencephaly

Figure .No.3: Meningomyelocoele

Figure .No.4: Occipital encephalocoele

Table 2: Internal anomalies seen in terminated and stillborn babies

S. no.	Type of anomalous foetus	Dysplastic kidney	Lobulated kidney	Adrenal gland agenesis	Asplenia	Hepatomegaly	Hypertrophy of Thymus
1	Anencephaly	-	-	+	-	+	-
2	Anencephaly	+	-	-	-	-	-

3	Anencephaly	-	-	-	-	-	-
4	Meningomyelocele	-	-	+	-	-	-
5	Meningomyelocoele	-	+	-	+	-	-
6	Meningomyelocoele	+	-	-	-	-	-
7	Occipital encephalocele	+	+	-	-	-	-
8	Meningocoele	-	-	-	-	-	-
9	Meningocoele	-	-	-	-	-	-
10	Skeletal Dysplasia	+	-	-	-	-	-
11	Skeletal Dysplasia	-	-	-	-	-	+
12	Skeletal Dysplasia	-	-	-	-	-	-
	Total	**4**	**2**	**2**	**1**	**1**	**1**

(+) - denotes congenital anomaly (-) - denotes normal

Figure No. 5: Agenesis of supra renal gland in meningomyolocoele

Figure No. 6: Bilateral lobulated kidney &Asplenia in meningomyolocoele

Figure No. 7: Huge hepatomegaly in anencephaly

Figure No. 8: Hypertrophy of thymus in skeletal dysplasia

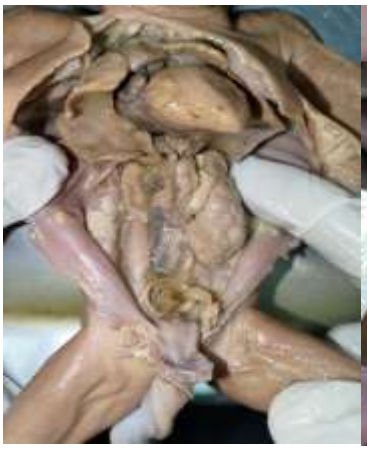

Figure No. 9: Left side lobulated and right side dysplastic kidney in occipital encephalocoele

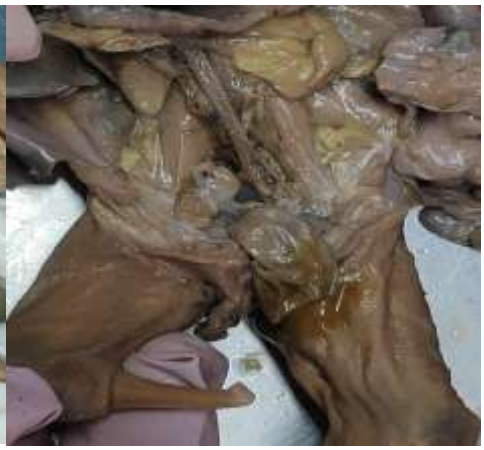

Figure No.10: Dysplastic kidney in skeletal dysplasia

4. Discussion

Foetal loss is a common clinical problem and the family needs to know the cause of the loss of a foetus. The future reproductive decision of the couple depends on the cause of the foetal loss that will predict the recurrence risk and may prevent similar losses. The investigations to be performed for analyzing foetal loss includes radiograph, chromosomal analysis, foetal autopsy, investigations for infections and genetic metabolic causes, histopathology of placenta and other foetal tissues as indicated.[8,9] Chromosomal analysis should be performed not only for major malformation but also in foetal hydrops, intrauterine growth retardation, oligohydramnios, macerated fetus and unexplained foetal loss. Skeletal radiograph helps in detecting skeletal dysplasia both prenatally and following termination [10,11]. Detecting anomalies and malformation prenatally helps in reducing the risk of recurrence which could be otherwise upto 25%. Disproportionate shortening of the limbs becomes more apparent during childhood. Although all appendicular skeletal elements are short, the distal and middle segments are generally more severely affected than the proximal segment[12]. Acromesomelic dysplasia is characterized by markedly short hands and feet, dwarfism to variable degree and characteristic face with narrow palpebral fissures, short stubby nose and averted nostrils[13].

In our study male and female ratio is nearly 2: 1. The most common anomalies were related to

urinary and gastrointestinal system. The urogenital abnormality in this study is very well correlated with the study of sivasankaranayak [14] & A.G. Tomatir et al [15].

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 diagnosis. In such cases the storage of foetal samples for possible future genetic analysis provides the hope of an accurate diagnosis 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. 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.

5. Conclusion

In the present study the incidence of congenital anomalies was higher in stillborn with mother having bad obstetric history and urinary system anomalies is the most common internal congenital malformation. Early detection and termination of congenital anomaly will reduce the birth of babies with congenital anomalies. It will also decrease the economic burden, psychological trauma to the parents and family.

Even through the prenatal ultrasonography reasonably predicts the malformations. Foetal autopsy is essential to look for additional internal malformations. In this study foetal autopsy helps in confirming the diagnosis of congenital malformations detected by antenatal ultra sound findings and identifying the additional internal malformations and which will help in genetic counseling of the couple.

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