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STUDY OF CONGENITAL ANOMALIES IN STILLBORN FETUSES THROUGH AUTOPSY

Anatomy

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ABSTRACT

Background And Aims: Congenital anomalies may be defined in terms of physical structure as a malformation which are present at the time of birth or functional disturbance as a defect in which there is sufficient disturbance in the usual number, size, shape and location or inherent character of any part, organ, cell or its constituent. Therefore, congenital anomalies are alteration of normal anatomic structure present at the time of birth and are of social, diagnostic and clinical importance. The aim of the present study is to categorize the congenital anomalies based on the system of involvement through autopsy with a focus on sex ratio. **Materials And Methods:** This study consists of 36 still born fetuses and the fetuses are collected from the Department of Obstetrics and Gynecology, JSS Hospital, Mysore. Fetus is fixed in 10% formalin and formalin is injected in thoracic cavity, abdominal cavity and cranial cavity for fixation of the organs. The autopsies were carried out as per guidelines provided by standard fetal autopsy protocol. **Results:** The occurrence of Congenital Anomalies was more common in males than females. The pattern of Congenital Anomalies included CNS, Digestive system, urinary system and circulatory system. **Conclusion:** Even though the prenatal ultrasound reasonably predicts the malformations, fetal autopsy is essential to look for additional malformations. This study confirms the utility of fetal autopsy in identifying the cause of fetal loss which will help in counseling of the couple for future family planning.

KEYWORDS

Congenital anomalies; Autopsy; Still born

INTRODUCTION:

Congenital anomalies may be defined in terms of physical structure as a malformation which are present at the time of birth or functional disturbance as a defect in which there is sufficient disturbance in the usual number, size, shape and location or inherent character of any part, organ, cell or its constituent. Therefore, congenital anomalies are alteration of normal anatomic structure present at the time of birth and are of social, diagnostic and clinical importance.[1]

There are four main types of congenital anomalies which include malformations, disruptions, deformations and dysplasias. Malformations represent morphologic defects resulting from intrinsically abnormal developmental process and occur early in embryogenesis.

Disruptions are breakdown of or interference with an originally normal developmental process and may occur at any time during gestation. Deformation can result from mechanical interference with the normal growth, functioning or positioning of the fetus in utero. Dysplasias result in abnormal structure because the tissues from which individual structures formed are abnormal.[2]

The term "Autopsy" was derived from Greek word "autopsia" which means "to see one oneself" (autos – "oneself" and opsia – "eye"). Perinatal autopsy provides valuable information which facilitates emotional and psychological recovery, exact diagnosis for proper counseling and future pregnancy planning. Once a couple faces a fetal demise there are always concerns about the cause of death and its recurrence risk. Perinatal autopsy remains the gold standard in the investigation of perinatal death. Sometimes the antenatal ultrasound examinations fail to identify the cause of death and in such cases autopsy is must.[3]

The International Classification of Diseases, 10th revision (ICD-10) defines a fetal death as "death prior to the complete expulsion or extraction from its mother of a product of conception, irrespective of the duration of pregnancy; the death is indicated by the fact that after such separation the fetus does not breathe or show any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles" without specification of the duration of pregnancy. ICD classifies late fetal deaths (greater than 1000gms or after 28 weeks) and early fetal deaths (500 to 1000gms or 22-28 weeks). [4] Even though the prenatal ultrasound reasonably predicts the malformations, fetal autopsy is essential to look for additional malformations. This study confirms the utility of fetal autopsy in identifying the cause of fetal loss which will help in counseling of the couple for future family planning. Hence, this study becomes essential because some associated malformations can be missed or are undetectable on ultrasound.

MATERIALS AND METHODS:

This study consists of 36 still born fetuses (22 male and 14 female). The fetuses were collected from the Department of Obstetrics and Gynaecology, JSS Hospital, Mysore. This study was approved by Institutional Ethics Committee of JSS Medical College, Mysuru. Written consents were obtained from the parents of all the still born fetuses before conducting the autopsy.

Inclusion criteria: All still born fetuses at or above 24 weeks and fetuses whose relatives gave informed consent were included.

Exclusion criteria: Fetuses with gestational age less than 24 weeks and autolysed fetuses were excluded.

Fetuses were fixed in 10% formalin and formalin was injected in thoracic cavity, abdominal cavity and cranial cavity for fixation of the organs. The autopsies were performed as per guidelines provided by standard fetal autopsy protocol. A longitudinal incision was taken from the suprasternal notch up to the pubic symphysis followed by a horizontal incision to retract the skin flap. The thoracic and abdominal cavities were opened and any deviation from the normal anatomy were noted and photographed.

RESULTS:

Below is the tabular representation of system wise classification of congenital anomalies [Table 1]. Central nervous system anomalies (27.7%) are the commonest anomalies observed in the present study. Cardiovascular system (13.9%), Gastrointestinal system (13.9%), Genitourinary system (13.9%), Musculoskeletal system (13.9%) and miscellaneous cases (16.7%).

Table 1: System-wise classification of congenital anomalies

Serial No	Systems involved	Number	Percentage (%)
1	Cardiovascular system	5	13.9
2	Gastrointestinal system	5	13.9
3	Genitourinary system	5	13.9
4	Central Nervous System	10	27.7
5	Musculoskeletal system	5	13.9
6	Miscellaneous	6	16.7

A case of meningomyelocele was observed in 28 weeks male fetus (Figure 1) and another case of anencephaly was observed in 26 weeks male fetus (Figure 2).

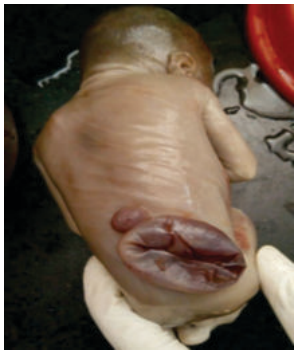

Figure 1: Meningomyelocele in 28 weeks male fetus

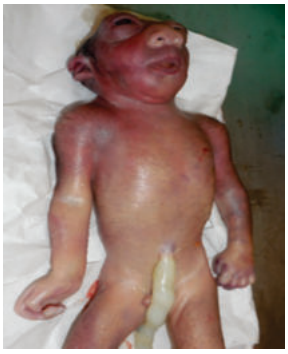

Figure 2: Anencephaly in 26 weeks male fetus

DISCUSSION:

Congenital Malformation is a physical, metabolic or anatomic defect which is apparent before birth, at birth, or detected during the first year of life. The evaluation of congenital malformations can be done by ultrasound, maternal serum analysis, etc., prenatally and by autopsy after the fetal death. 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, data pertaining to demography, socio-economic status and maternal health is helpful to pinpoint the factors the occurrence of the fetal loss.[5]

In a study of 150 fetal autopsies by Kanchan Kapoor et al, 87 (58%) specimens were induced abortions, and 63 (42%) spontaneous abortions and intrauterine deaths. In total, the incidence of congenital malformations was 104 (69%). Among 150 fetuses, 74 were males and 76 female fetuses; out of which the incidence of congenital malformations in males and females was 75% and 83% respectively. The types of congenital malformations were classified as central nervous system defects (CNS) in 49 (33%), gastrointestinal tract (GIT) disorders in 48 (32%), musuculoskeletal (MS) disorders in 31 (21%), genito-urinary (GU) in 25 (17%), and genetic disorders in 12 (8%). Multiple anomalies were present in 40 (27%) fetuses. Anencephaly was the most prevalent anomaly (29%).[5]

In this study of 100 perinatal autopsies by Andola US et al, 44 cases of congenital anomalies were encountered with M: F ratio of 1:1.5. The gestational age of most of the fetuses with congenital anomalies ranged from 35-39 weeks & birth weight range 350-1000g. The most common defects were of Central Nervous System, seen in 15 cases (34.09). The most common defect was Anencephaly with 11 cases (25%), which were accurately diagnosed with Ultrasonography. The next common anomalies were of Renal system accounting for 9 cases (20.45%) which included renal agenesis, polycystic kidney disease, Wilms tumor, obstructive uropathy and horseshoe kidney which was incidentally observed in a case of Hydrops Fetalis.[6]

There were 7 cases (15.91%) of multiple malformations not fitting into any specific diagnosis, 5 cases (11.36%) of syndromes diagnosed as Prune Belly syndrome, Meckel Gruber syndrome, Thanatophoric Dysplasia Type 1, Mermaid syndrome or Sirenomelia and OEIS complex (omphalocele, exstrophy of the bladder, imperforate anus, and spinal abnormalities/ myelomeningocele,), 3 cases (6.82%) of diaphragmatic hernia, 2 cases (4.54%) of congenital heart defects and 2 cases (4.54%) of congenital cystic adenomatoid malformations lung. A case of Infantile hemangioendothelioma liver was encountered.[6]

According to Padma S et al obtaining consent for autopsy was the major hindrance for screening more number of fetuses. Out of 102 autopsies, congenital anomalies were found in 28 fetuses (27%). The most common anomalies were that of central nervous system with Anencephaly being the commonest one. In one fetus we found Anencephaly associated with Spinabifida which was the only anomaly found in rest of the fetus with central nervous system anomalies. Digestive system was the next commonest one followed by urinary system. Circulatory system was the least common type. The incidence of these congenital anomalies was found to be more in male fetus on comparison to the females ($p < 0.01$).[7]

In a study of 100 still born fetuses by Ravikiran Ashok Gole et al, 54 were males and 46 were females. Obtaining the consent was the main hindrance which compelled us to restrict the sample size. Of the total cases, congenital anomalies were obtained in 36 cases, of which 17 were males and 19 females. Central nervous system was the commonest system involved followed by musculoskeletal system. During the most sensitive period of embryogenesis i.e. the 3rd - 8th weeks of gestation factors like genetic, environmental, teratogenic and infectious agents play important role for the origin of malformations. The significance of sex predominance is important as the dominance of a particular malformation helps to predict the occurrence and outcome. In this study, significant sex predominance was found in only anencephaly which was common in females. All the previous studies have found anencephaly to be common in females.[8]

In a study by Taksande et al, out of total 9386 deliveries, 9196 were live births and 192 were stillbirths. The sexwise distribution was 62% males and 38% females, giving an M:F ratio of 1.63:1, and there were 3 cases of ambiguous genitalia. Congenital malformations were seen more significantly in stillbirths ($P < 0.01$) as compared to live births, the frequency being 4.68% and 1.84%, respectively. Nine of the 179 malformed babies (5.02%) were still born. Prematurity, increased maternal age, increasing birth order and low birth weight were found to have a higher of congenital anomalies. Cardiovascular malformations were most common followed by musculoskeletal and genitourinary anomalies.[9]

In this retrospective study of 206 cases by Sankar VH et al, stresses the need of fetal autopsy in providing accurate genetic counseling. Autopsy facilities are not yet commonly available in India. There is a need to educate obstetricians about the need of fetal autopsy and placental histology for genetic counseling. Pediatricians with interest in malformation can take up the responsibility of establishment of fetal autopsy facilities in collaboration with a pathologist. The uptake of perinatal autopsy services depends on the awareness among obstetricians and pediatricians about its need and utility.[10]

CONCLUSION:

This study highlights the importance of fetal autopsy in providing accurate genetic counseling. There is a need to bring awareness among obstetricians about the need of fetal autopsy and placental histology for genetic counseling. Pediatricians with interest in malformation can take up the responsibility of establishment of fetal autopsy facilities in collaboration with a pathologist. The uptake of perinatal autopsy services depends on the awareness among obstetricians and pediatricians about its need and utility.

REFERENCES:

1. Parmar A, Rathod SP, Patel SV, Patel SM. A Study of Congenital Anomalies in Newborn. NJIRM. 2010 ; 1 (1) : 13-17.
2. Moh W, Graham JM, Wadhawan I, Sanchez-Lara PA. Extrinsic Factors Influencing Fetal Deformations and Intrauterine Growth Restriction. Journal of Pregnancy. Volume 2012 ; 1-11.
3. Pradhan R, Mondal S, Adhya S, Raychaudhuri. Perinatal Autopsy : A Study from India. J Indian Acad Forensic Med. 2013 ; 35 (1) : 10-13.
4. Lawn JE, Gravett MG, Nunes TM, Rubens CE, Stanton C et al. Global report on preterm birth and stillbirth (1 of 7): definitions, description of the burden and opportunities to improve data. BMC Pregnancy and Childbirth. 2010; 10 (Suppl 1): 1-22.
5. Kapoor K, Singh K, Sharma A, Singh B, Huria A, Kocchar S. Congenital anomalies in North Western Indian population – a fetal autopsy study. Eur J Anat. 2013; 17(3): 166-175.
6. Andola US, Anita AM, Ahuja M, Andola SK. Congenital Malformations in Perinatal Autopsies – A Study of 100 Cases. Journal of Clinical and Diagnostic Research. 2012; 6(10): 1726-1730.
7. Padma S, Ramakrishna D, Jijiya BP, Ramana PV. Pattern of distribution of congenital anomalies in stillborn: A hospital based prospective study. International Journal of Pharma and Bio Sciences. 2011; 2(2): 604-610.
8. Gole RA, Meshram PM, Hattangdi SS. Congenital malformations in still born fetuses. International Journal of Recent Trends in Science and Technology. 2014; 11(3): 412-414.
9. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian Journal of Human Genetics. 2010; 16(3): 159-163.
10. Shankar VH and Phadke SR. Clinical utility of fetal autopsy and comparison with prenatal ultrasound findings. Journal of Perinatalogy. 2006; 26: 224-229.