Original Research Article

A hospital based fetal autopsy study of pattern of congenital anomalies at a tertiary hospital

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Abstract

Background: Perinatal autopsy either confirms or refute the ultrasound diagnosis and complements USG examination in making a precise diagnosis. Present study was aimed to study pattern of congenital anomalies based on fetal autopsy at a tertiary hospital. **Material and Methods:** Present study was hospital based, observational and descriptive study, conducted in autopsies of fetuses with gestational age > 20 weeks OR birth weight > 500 gms. and autopsies of neonates within 7 days of post-natal life. Malformations were classified into organ systems according to World Health Organization (WHO) recommendations. **Results:** In present study 90 fetuses/neonates underwent autopsy for various congenital anomalies. Majority of mothers were from 21-30 years age group (53.33 %), were primigravida (37.78%). Majority of fetuses/neonates those underwent autopsy were stillbirth delivered after 28 weeks (61.11 %). Male (56.67 %) fetuses/neonates were more than female (41.11%) and 2 had ambiguous genitalia. In present study various Associated/ high risk factors / findings noted were abnormalities of AFI (40 %), Hypothyroidism (10 %), H/O of anomalous baby (7.78 %), Consanguinity (6.67 %), Uncontrolled diabetes mellitus (4.44 %) and h/o fever in first trimester (1.11 %). In present study major systems involved were central nervous system (28.89%), urinary system (27.78%), cardiovascular system (25.56%) and musculoskeletal system (21.11%). Out of 90 fetuses/neonates underwent autopsy, 86 had antenatal USG. Among them in majority of cases USG findings were confirmed on autopsy (59.3 %), while USG findings confirmed on autopsy as well additional findings were noted in 29 cases (33.72 %). In 6 cases there was change in diagnosis (6.98 %). **Conclusion:** There is a good correlation between prenatal ultrasound scanning and autopsy findings. Clinically, autopsy is important for genetic counselling and planning of next pregnancy.

Keywords: Autopsy, Congenital abnormalities, Prenatal diagnosis, Perinatal mortality, Syndrome

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INTRODUCTION

A congenital anomaly is defined in terms of physical structure as a malformation, an abnormality of physical structure or form usually found at birth or during the first few weeks of life.[1] Congenital malformations remain a common cause of perinatal deaths accounting for 10-15% in developing countries like India.[2] There are more than 4000 different birth defects, in which some of the defect is treated and cured and other are not treated, leading to death in the first year of life. An estimated 276,000 babies die within 4 weeks of birth every year, worldwide, from congenital anomalies.[3] Congenital malformations are the most common reason for perinatal deaths, with autopsy having an additive role to prenatal and genetic evaluations and providing foresight for planning a subsequent pregnancy.[4,5] Birth defects may range from the very minor or easily treatable abnormality with excellent functional prognosis to those that are complex and difficult to manage having very poor outcome. Ultrasonographic (USG) examination can accurately diagnose many lesions but has its own limitation in detecting soft-tissue lesions such as malrotation of intestines, asplenia/polysplenia, and pulmonary hypoplasia, and the success rates of ultrasound also depend on the equipment used and expertise of the

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sonologist.[4] Perinatal autopsy either confirms or refute the ultrasound diagnosis and complements USG examination in making a precise diagnosis. Present study was aimed to study pattern of congenital anomalies based on fetal autopsy at a tertiary hospital.

MATERIAL AND METHODS

Present study was hospital based, observational and descriptive study, conducted in department of forensic medicine and toxicology, at Konaseema Institute of Medical Sciences and Research Foundation, Amalapuram, Study approval was obtained from institutional ethical committee.

Inclusion criteria: Autopsies of fetuses with gestational age > 20 weeks OR birth weight > 500 gms. and autopsies of neonates within 7 days of post-natal life.

Exclusion criteria: Autolysed dead fetuses or neonates

Study was explained and a written informed consent for the autopsy was taken from the parents/relatives. Relevant antenatal/past history (maternal history), clinical details (antenatal investigations), delivery details were collected and entered in a predesigned proforma for detection of congenital malformations in fetal and early neonatal autopsies. Prior to autopsy, radiographs of abnormal features were taken whenever required. Prior to autopsy, morphometric examination was done followed by autopsy as per standard operative procedures. During autopsy procedure, photographs were taken for unusual gross findings. Sections from each organ were sent histopathological examination. Autopsy findings were compared with antenatal ultrasound findings if available. Malformations were classified into organ systems according to World Health Organization (WHO) recommendations.[5] Data was collected and compiled using Microsoft Excel, analysed using SPSS 23.0 and statistical analysis was done using descriptive statistics.

RESULTS

In present study 90 fetuses/neonates underwent autopsy for various congenital anomalies. Majority of mothers were from 21-30 years age group (53.33 %), were primigravida (37.78%). Majority of fetuses/neonates those underwent autopsy were stillbirth delivered after 28 weeks (61.11 %). Male (56.67 %) fetuses/neonates were more than female (41.11%) and 2 had ambiguous genitalia.

Table 1: General characteristics

Characteristic	No of anomalous babies (n=57)	Percentage
Maternal age (in years)		
≤20	9	10.00%
21-30	48	53.33%
31-35	22	24.44%
>35	11	12.22%
Gravida status		0.00%
1	34	37.78%
2	27	30.00%
3	22	24.44%
≥ 4	7	7.78%
Fetal age		0.00%
< 28 weeks	18	20.00%
>28 weeks	55	61.11%
Early neonatal period (< 7 days)	17	18.89%
Gender		
Male	51	56.67%
Female	37	41.11%
Ambiguous	2	2.22%

In present study various Associated/ high risk factors / findings noted were abnormalities of AFI (40 %), Hypothyroidism (10 %), H/O of anomalous baby (7.78 %), Consanguinity (6.67 %), Uncontrolled diabetes mellitus (4.44 %) and h/o fever in first trimester (1.11 %).

Table 2: Associated factors

Associated factors		0.00
Abnormalities of AFI	36	40.00%
Hypothyroidism	9	10.00%
H/O of anomalous baby	7	7.78%
Consanguinity	6	6.67%
Uncontrolled diabetes mellitus	4	4.44%
h/o fever in first trimester	1	1.11%

In present study major systems involved were central nervous system (28.89%), urinary system (27.78%), cardiovascular system (25.56%) and musculoskeletal system (21.11%). Central Nervous system (28.89%) anomalies were most common and malformations noted were Spina Bifida, Anencephaly, Hydrocephalus, Corpus Callosal Agenesis, Meningomyelocele, Craniofacial Rashischisis and Holoprosencephaly. Second most common system with anomalies were urinary system (27.78%) and malformations noted were Renal Agenesis, Polycystic kidneys, Dysplastic kidneys. Other systems with congenital anomalies were cardiovascular system (25.56%) and various malformations noted were Fallot's tetralogy, Ventricular Septal Defect, Atrial Septal Defect, Ventriculomegaly, Transposition of great vessels, Abnormal aorto-pulmonary communication and Hypoplastic heart syndrome. Musculoskeletal system (21.11%) malformations were CTEV, Limb deformity, Diaphragmatic hernia, Omphalocele, Imperforate Anus, Rocker Bottom foot, Arthrogryposis. Respiratory system (5.26 %) malformations in study cases were congenital cystic adenomatoid malformation and bilateral hypoplastic lungs.

Less common systems identified were Eye, Ear, Face and Neck system (6.67%), Lymphatic system - Cystic Hygroma (5.56%) and Ambiguous genitalia (2.22%). Syndromes identified (12.22%) were Down's syndrome, Edward's syndrome, VACTERAL- H syndrome, Syringomyelia, Limb body wall complex and Potter's syndrome.

Table 3: System wise distribution of Congenital Malformations

System affected in congenital malformations*	No of anomalous babies (n=90)	Percentage
Central Nervous system	26	28.89%
Spina Bifida	15	
Anencephaly	11	
Hydrocephalus	8	
Corpus Callosal Agenesis	6	
Meningomyelocele	5	
Craniofacial Rashischisis	2	
Holoprosencephaly	2	
Urinary system	25	27.78%
Renal Agenesis	12	
Polycystic kidneys	11	
Dysplastic kidneys	6	
Cardiovascular system	23	25.56%
Fallot's tetralogy	14	
Ventricular Septal Defect	12	
Atrial Septal Defect	11	
Ventriculomegaly	6	
Transposition of great vessels	4	
Abnormal aorto-pulmonary communication	2	
Hypoplastic heart syndrome	2	
Musculoskeletal system	19	21.11%
CTEV	7	
Limb deformity	7	
Diaphragmatic hernia	5	
Omphalocele	4	
Imperforate Anus	3	
Rocker Bottom foot	2	
Arthrogryposis	1	
Syndromes identified	11	12.22%
Down's syndrome	3	
Edward's syndrome	2	
VACTERAL- H syndrome	2	
Syringomyelia	2	
Limb body wall complex	1	
Potter's syndrome	1	
Eye, Ear, Face and Neck system	6	6.67%
Dysmorphic facial features	4	
Cleft Lip, Cleft Palate	2	
Bilateral Cataract	1	

Respiratory system	5	5.56%
Congenital Cystic Adenomatoid Malformation	4	
Bilateral Hypoplastic lungs	1	
Lymphatic system - Cystic Hygroma	5	5.56%
Ambiguous genitalia	2	2.22%

(*one or more systems involvement was noted in few cases)

Out of 90 fetuses/neonates underwent autopsy, 86 had antenatal USG. Among them in majority of cases USG findings were confirmed on autopsy (59.3 %), while USG findings confirmed on autopsy as well additional findings were noted in 29 cases (33.72 %). In 6 cases there was change in diagnosis (6.98 %).

Table 4: Correlation of Antenatal USG findings with Autopsy findings.

Correlation of USG findings and autopsy findings	Cases (n=86)	Percentage (%)
USG findings confirmed on autopsy	80	93.02%
• No change in diagnosis	51	59.30%
• Additional findings noted in the autopsy	29	33.72%
Changes in Diagnosis	6	6.98%

DISCUSSION

The key objectives of autopsy examination are identification of cause(s) of death, elucidation of pathogenic mechanism and quality control of clinical mechanism.[6] Autopsy is mandatory in congenital malformations, recurrent fetal death-after the obstetrician has investigated thoroughly for a cause and found none, hydrops fetalis-all cases of non immune hydrops, cases of IUGR where no cause is detectable, suspected infections, any Unexplained event e.g. failed resuscitation.[7] Shirke R *et al.*,[8] studied 2242 neonates, the prevalence of congenital malformations was 8.3%. Central nervous system and cardiovascular, musculoskeletal, and genitourinary systems were found to be most commonly involved. Higher prevalence of malformation in the babies born to mothers aged over 35 years reported by Suguna Bai *et al.*,[9] whereas Dutta and Chaturvedi[10] documented statistically insignificant association of increased maternal age and congenital anomalies. In present study, a majority of malformed babies were born to mothers aged less than 35 years. Arbil Açikalin *et al.*,[11] conducted 2150 autopsies performed for perinatal deaths. A pathology was detected in 1619 of 2150 (73.3%) autopsies. Congenital malformations were the most common diagnosis with 68.2%. Neural tube defects and central nervous system malformations were the most frequent system malformation in 28.8% of cases, followed by the urogenital system (11.4%) and musculoskeletal system (8.3%), respectively. Malformation syndromes including multisystem anomalies were defined in 109 cases (9.3%). In study by Anne GC *et al.*,[12] prevalence of congenital anomalies was 12.5 per 1000 live births, with musculoskeletal disorders being the commonest, followed by craniovertebral anomalies. The prevalence of anomalies over the past 10 years has not shown any significant change. The high prevalence of neural tube defects indicates the need for periconceptional folic acid supplementation and early detection of anomalies, which would help in timely management. Detection of musculoskeletal anomalies would help in counseling patients antenatally. Shailaja P *et al.*,[13] studied 23 fetal autopsies, among 14 male (60.86%) and 8 female (34.78%) fetuses, and in one case (4.34%) gender could not be identified. 19 cases (82.60 %) were < 28 weeks of gestation. Medical termination of pregnancy was done in 13 cases (56.52 %) whereas, 10 patients (43.47 %) had spontaneous intrauterine death of the fetus. Ultrasound scanning was done in 15 cases (65.21 %). In 13 cases (86.66 %) the ultrasound and autopsy findings were correlating whereas in two cases (13.33 %) there were findings on imaging study which could not be identified on autopsy. Ultrasound was not done in 8 cases (34.78 %) out of which 5 cases (62.5 %) showed findings on autopsy which could have led to the fetal demise. 12 cases (52.17 %) were referral cases which had come from other hospitals. Genetic studies were done in 6 cases (26.08 %) in the form of parental karyotyping and cord blood could be tested in only one case. Andola US *et al.*,[14] studied 100 perinatal autopsies, 44 cases were congenital anomalies with M:F = 1:1.5. Majority of the fetuses with congenital malformations (36.36%) were therapeutically terminated, Central nervous system malformations being the commonest indication. The most common timing of therapeutic termination being 20 -24weeks. Congenital malformations were common between 35-39 weeks gestational age and birth weight range 350- 1000g. The malformations involving the central nervous system were commonest (34.09%), followed by renal anomalies (20.45%) and multiple malformations (15.91%). Autopsy confirmed the prenatal ultrasound findings in 50% of the cases, added to diagnosis in 29.54%, while it completely changed the primary diagnosis in 9.09% of the cases. Bhide P *et al.*,[15] followed a cohort of 2107 pregnant women till outcome which was miscarriage, termination of

pregnancy, live or stillbirth, neonatal and post-neonatal mortality. Among 1822 births, the total prevalence of major congenital anomalies was 230.51 (170.99±310.11) per 10000 births. Congenital heart defects were the most commonly reported anomalies (65.86 per 10000 births), followed by neural tube defects (27.44 per 10000 births). In this cohort, congenital anomalies were the second largest cause of neonatal deaths. The congenital anomaly prenatal diagnosis prevalence was 10.98 per 1000 births and the congenital anomaly termination of pregnancy rate was 4.39 per 1000 births. There are a few factors that may contribute to increased incidence of anomalies in developing countries like inadequate periconceptional folic acid, consanguineous marriage,[16] increasing age group, low birth weight or intake of teratogenic drugs in pregnancy.[17,18] Despite advances in imaging such as antenatal ultrasonography and serology, perinatal autopsy is superior and continues to play an important role in diagnosing congenital malformations. Fetal and early neonatal autopsy in cases of congenital malformations not only confirms but also provides additional information such as estimating the risk of recurrence, for genetic counseling and is also helpful in counseling the parents regarding prevention of similar congenital malformations in future pregnancies. During the last few years, modern cross-sectional imaging techniques have pioneered forensic medicine. Magnetic resonance imaging and especially multiline computed tomography are becoming increasingly implemented into post-mortem examinations. these non-invasive techniques can augment and even partially replace a traditional autopsy.[19]

CONCLUSION

There is a good correlation between prenatal ultrasound scanning and autopsy findings. But, Fetal autopsy plays an important role in arriving at the final diagnosis and detecting the cause of death as well as can demonstrate more accurately congenital malformations and unsuspected cord abnormalities. Clinically, autopsy is important for genetic counselling and planning of next pregnancy.

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