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AUTOSPY STUDY TO DETERMINE FETAL ANOMALY: A RETROSPECTIVE COHORT STUDY

R M POTEKAR[1], JAVALGI P ANITA[*2] AND B R YELIKAR[3]

1.Professor , Department of Pathology , BLDEU's Shri B M Patil Medical College, Bijapur.
2. Assistant Professor , Department of Pathology , BLDEU's Shri B M Patil Medical College, Bijapur
3. Professor & Head , Department of Pathology , BLDEU's Shri B M Patil Medical College, Bijapur

ABSTRACT

Congenital anomalies have become an important cause of perinatal mortality and morbidity in both developing and developed countries. Present study was done to know the frequencies of congenital anomalies, its association with maternal diseases, maternal age and co-relation with ultrasonographic findings.Total 54 autopsy cases were studied in 6 year span from January 2007- December 2012.Consent for autopsy was obtained from either of the parent after explaining the need. Each foetus was examined according to predetermined protocol which included ultrasound diagnosis, photograph, whole body X ray (when required), external and internal examination.Out of 54 fetal autopsies, 35(64.8%) cases had congenital anomaly, 10cases (18.5%) had shown placental abnormality and 6cases (11.1%) had shown umbilical abnormality. Congenital anomalies were common between gestational age 35-39weeks and birth weight range 250-1000grams. This study highlights the importance of fetal autopsy in confirming the prenatal diagnosis

KEY WORDS: autopsy / prenatal diagnosis/ fetal anomaly/maternal diseases.

JAVALGI P ANITA
Assistant Professor , Department of Pathology , BLDEU's
Shri B M Patil Medical College, Bijapur

INTRODUCTION

Congenital anomalies have become an important cause of perinatal mortality and morbidity in both developing and developed countries[1]. It accounts for 10-15% of perinatal death in developing countries [2] and 25-30% in developed countries[3]. Unlike situation in developed countries, the leading cause for infant morbidity and mortality in developing countries are low birth weight, prematurity, sepsis and infections[4]. The Abortion Act 1967 allows termination of pregnancy if there is substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously disabled. During the past decade new screening and diagnostic tests have been introduced that allow earlier and more accurate diagnosis of fetal anomaly[5'6]. After termination of pregnancy most parents will want to know if the prenatal prediction was accurate and what the implications are for future pregnancies. This is particularly true when the prenatal diagnosis is based on ultrasound scan findings only. Such information may be obtained from the autopsy examination[7]. Hence present study was done to know the frequencies of congenital anomalies, its association with maternal diseases, maternal age and co-relation with ultrasonographic findings.

METHODOLOGY

All terminated pregnancies from 12-40weeks due to abnormal prenatal findings, IUD and deaths within the 7completed days after delivery were included in the study. Newborns with more than 7days of life and then expired were excluded from the study. Total 54 autopsy cases were studied in 6 year span from January 2007- December 2012. Consent for autopsy was obtained from either of the parent after explaining the need. Each foetus was examined according to predetermined protocol which included ultrasound diagnosis, photograph, whole body X ray, external and internal examination. The autopsy protocol included the removal of thoracic, cervical, abdominal and pelvic organs en block and subsequently dissected into organ blocks[8]. The placenta, fetal membranes and umbilical cord were studied in all the cases. Histological sections were taken from lung, liver, kidney, thymus, brain, placenta and umbilical cord. In cases where the antenatal ultrasonography diagnosis was available, were compared with the postnatal autopsy findings.

RESULTS

Total autopsy cases studied were 54 from 2007-2012 Maternal age group were predominantly between 20-30yrs, 2 cases were below 20yrs youngest was 15yrs, and 6cases were above 30yrs eldest was 38yrs old lady. Out of 54 fetal autopsies, 35(64.8%) cases had congenital anomaly and 19(35.2%) cases had no abnormality, 10cases (18.5%) had shown placental abnormality out of 54 placental examination and 6cases (11.1%) had shown umbilical abnormality (Table I). Among 54 cases, 9 were therapeutic termination, 27were still birth and 15 were intra-uterine death and 3 were perinatal death.

TABLE I
DISTRIBUTION OF SPECIFIC PATHOLOGY

Type of pathology	No of cases with abnormality	No of cases With no abnormality
Foetal anomaly	35 (64.8%)	19 (35.2%)
Placental abnormality	10 (18.5%)	44 (81.6%)
Umbilical abnormality	6 (11.1%)	48 (88.9%)
Maternal diseases	44 (81.4%)	10 (18.6%)

Congenital anomalies were common between gestational age 35-39weeks following 25-29weeks and least in perinatal deaths. Birth weight ranging between 250-1000grams (42.59%) were showing most congenital anomalies. The most common congenital anomaly detected was musculoskeletal defect, central nervous system and respiratory system followed by uro-genital anomalies (Table II). We observed four interesting cases which included Body stalk anomaly (fig 1), De George syndrome(fig 2), Ellis van crevald syndrome and OEIS (omphalocele extrophy imperforate anus spinal defects)

Table II
system wise distribution of congenital anomalies

Defects	No of cases (n=35)
Central nervous defects	**9 CASES**
• Meningocele	1
• Anencephaly + meningocele	1
• Spina bifida + Meningomyelocele	2
• Spina bifida + meningocele	1
• Microcephaly + Arnold corpus callosum	1
• Congenital hydrocephalus	2
• Anencephaly	1
Musculoskeletal defects	**10 CASES**
• Polydactyly	4
• Syndactyly	3
• Absence of monodactyly	2
• unspecified	1
Urogenital defects	**7 CASES**
• Renal cystic lesions	2
• renal agenesis	2
• cryptorchidism	2
• hydrocele	1
Respiratory system defect	**9 CASES**
• diaphragmatic hernia	2
• hypoplastic lungs	5
• cystic adenomatoid malformation	2

Most common placental abnormality noted was hypertensive changes and infarction which were associated with pre-eclampsia and eclampsia conditions. There were 6 umbilical cord with single artery and these umbilical cord anomaly was noted with the all 4 syndromes and 2case of CNS anomaly. Maternal conditions associated with congenital anomalies was anemia as commonest condition , followed by pre-eclampsia, polyhydriominos, oligohydrimonis and two cases had severe infection diagnoses as cerebral malaria and positive for CMV and HSV infection (Table III)

Table III
Maternal diseases associated with congenital anomalies

Diseases	No of cases (44 CASES)
Anemia	25
Infection	
• Cerebral malaria	1
• CMV & HSV positive	1
Pre-elampsia & eclampsia	7
Polyhydrominos	6
Oligohydrominos	4

Ultrasonography findings were not obtained in all cases as their were cases without ANC check up. In only 36 cases sonography findings were available as shown in table IV which were correlated with autopsy findings.

Table IV
USG findings including placental abnormalities and fetal anomaly.

USG FINDINGS	NO OF CASES (36)
Placental abnormality	**5**
• Central placental previa	1
• Abruption placenta	4
Amniotic fluid	**10**
• Hydrominos	6
• Oligohydrominos	4
Fetal abnormalities	**21**

DISCUSSION

In an unselected population over a 10 year period there has been a decline in the number of fetal autopsies despite an increase in the number of terminations of pregnancy after prenatal suspicion of fetal abnormality[9]. A fall in autopsy rates has also been reported by other studies[7]. Hence present study was an attempt to study various parameters responsible for still birth, IUD or therapeutic cause for termination and significance of autopsy in making final diagnosis and to know the cause for termination of pregnancies. Fetal autopsy significantly contributes to the diagnosis of Intrauterine fetal death and congenital anomalies are a major cause of perinatal death[10]. In the present study of 54 perinatal autopsies, 35(64.8%) cases had congenital anomaly, with M:F ratio of 1:1.5. The most common mode of death was therapeutic termination of pregnancy (36.36%) and CNS malformations were most common indication for the same. The gestational age of most of the foetuses with congenital anomalies ranged from 35-39 wks & birth weight range 250-1000g. The most common defects were of musculoskeletal defect and Central Nervous System which correlates with the study of Kaiser et al[11], and Tomatir et al[12] and Andola et al[2] [Table V].

Table V
comparative analysis of congenital defects .

S No.	Authors	Year	Number/Total	Percentage (%)
1.	Kaiser et al	2000	45/121	37
2.	Sankar and Phadke	2006	60/81	74.2
3.	Grover N et al	2007	72/180	40
4.	Tomatir et al	2009	57/183	31.1
5.	Andola et al	2012	15/44	34.09
6.	Present study	2013	35/54	64.8

Present study observed respiratory disorders as second most common anomaly followed by uro-genital anomalies in contrast with studies done by Andola et al and Phadke et al where uro-genital anomalies were second common malformations[2,4]. Rare syndromes were encountered like Limb body wall complex /Body stalk anomaly refers to a rare complicated polymalformative fetal malformation syndrome of uncertain etiology. First described by Van Allen et al in 1987.There are very few cases reported in literature which is characterised by short umbilical cord and abdominal wall defect with malformed organs[13]. The DiGeorge Syndrome was first described in 1968 as a primary immunodeficiency resulting from the abnormal development of the third and fourth pharyngeal pouches during embryonic life. It is characterized by hypocalcemia due to hypoparathyroidism, heart defects, and thymic hypoplasia or aplasia[14]. OEIS complex, involving omphalocele, exstrophy of the bladder, imperforate anus, and spinal abnormalities/ myelomeningocele, is a rare association with incidence of 1 in 200 000 to 1 in 250 000 births. The present case had ambiguous genitilia and club foot which are

described as associated features [15]. There are very few studies carried out to evaluate maternal conditions influencing fetal anomaly , present study was an attempt to know the underlying relation between maternal conditions and fetal anomaly and observed that maternal infection and amniotic fluid abnormalities like polyhydriomonis or oligohydrominosis were associated with uro-geniatal anamolies. In the present study, autopsy diagnosis confirmed Ultrasound findings in 21cases (38.88%). The findings in this study were similar to those of Sankar and Phadke & Yeo et al[16,] Parents are very apprehensive and it is difficult decision to terminate the pregnancy due to congenital anomaly and also they are curious to know how it is going to affect the subsequent pregnancies Studies have shown that when the final prenatal diagnosis was made by ultrasound scan, in 27% of cases the information from the autopsy examination led to a refinement of the risk of recurrence, and in 8% this was increased to a one in four risk, these data may be of particular value to parents, and to those counselling them, when they are faced with a decision about whether to proceed with fetal autopsy after termination of pregnancy for suspected fetal anomaly[4].

CONCLUSION

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. 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.

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