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Congenital Anomalies – An Ultrasonographic and Autopsy Correlated Study

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ABSTRACT

Congenital anomalies became a crucial explanation for perinatal mortality in developed countries and developing countries like India. Worldwide surveys showed that birth prevalence of birth defect varies greatly from country to country which can be explained by social, racial, ecological and economical influences. Birth defect, leading explanation for foetal loss contribute significantly to preterm birth and childhood and adult morbidity, supported a study by Indian Academy of Paediatrics (IAP) reports that second major explanation for Infant deaths in Kerala is congenital anomalies (28%). Present Kerala's Infant death rate is 10per1000 livebirths in 2017(0.00%) and Maternal death rate 40/1,00,000 live births in 2019 getting to be decreased MMR to thirty in 2020 and twenty in 2030. Purpose of this study is to research the potential value of antenatal scan from six to twenty weeks for the assessment and diagnosis of congenital anomalies with a view to stop the birth of such babies, to work out the pattern of the congenital anomalies, to categorize them system wise and to work out the sex ratio of foetuses with congenital anomalies. Fifty cases of detected birth defect cases by Ultrasound scan and specimen of Male and Female foetuses obtained from medical termination of pregnancy of six to twenty weeks. A Proforma showing maternal datas, autopsy findings and Ultrasound findings. Anomalies were classified system wise into four categories in correlation with Ultrasound and Autopsy findings and foetuses with multiple congenital anomalies of varied systems. In System wise classification, commonest anomalies constitute Central systemanervosum (40%). Second commonest anomalies were of Urogenital system (36%). Category wise classification of congenital anomalies. Category A (full agreement between Ultrasound and Autopsy) had 44%. Category B (Autopsy confirmed all Ultrasound findings, but provided additional information of congenital anomalies) includes 38%. Category C (US findings were only partially demonstrated at Foetal autopsy; some anomalies revealed at US weren't verified at autopsy) includes 14% and Category D (Total disagreement between US and autopsy findings includes 4%; malformation whose diagnostic technique led to termination of pregnancy wasn't demonstrated at autopsy. Foetuses with multiple congenital anomalies two of the cases showed Meckel Gruber Syndrome includes a triad of Central systemanervosum malformation (Occipital encephalocoele), Polydactyly (postaxial) and Renal cystic dysplasia.

Keywords:
Indian Academy of Paediatrics, Perinatal mortality, Diagnostic procedure , Polydactyly

Introduction

The term congenital anomaly refers to an anatomic abnormality that's present at the time of birth. The term birth defect is typically used synonymously with Congenital anomaly, but birth defect is additionally want to incorporate metabolic and functional abnormalities.

The two broad subsets of congenital abnormalities are major and minor anomalies. Minor anomalies are those without surgical or cosmetic significance. Eg.Curvature (Clinodactyly) of the fifth finger or Partial webbing between the second and third toes.Major anomalies refers to people who require medical / surgical intervention / have cosmetic significance. These anomalies occur in 2-3% of all newborns and include such problems as Congenital heart defects, Cleft lip and Hydrocephalus. Although Prenatal Ultrasonography doesn't reveal all anomalies, the

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sonographic identification of the many anomalies enables further antenatal evaluation and opens the door to Parental Counseling.

Malformation denotes a structural defect that happens as a consequence of abnormal morphogenesis during the embryonic development of an organ / a part of an organ. As such, a cardiac defect thanks to an abnormal gene would be termed a malformation, whereas an equivalent cardiac defect resulting from a teratogenic agent eg. Rubella virus would be most properly termed an disruption. A foetal echocardiogram is usually indicated when an anomaly is discovered sonographically in another organ system.

Syndrome may be a pattern of multiple anomalies that can't be ascribed to one initiating event during a cascade but that's though nevertheless to be the consequence of one causative factor. All of the malformations one might encounter in Mongolism .Eg.Cardiac anomaly.

Association refers to the nonrandom occurrence of multiple anomalies as seen in additional than one individual and not known to represent a sequence / a syndrome.

At present Kerala's infant deathrate Rate 10 per 1000 live births in 2017(0.00%) and Maternal deathrate 40/1,00,000 live births in 2019. Purpose of this study is to research the potential value of antenatal scan done during intrauterine life from six to twenty weeks for the assessment and diagnosis of Congenital anomalies with a view to

1. Prevent the birth of such babies.
2. to work out the pattern of the Congenital anomalies
3. to work out the sex ratios of foetuses with Congenital anomalies.

Teratologic factors are termed in five major groups:

1. Physical (X-rays radiation)
2. Chemicals (drugs employed by Pregnant women)
3. Nutritional (hypervitaminosis, hypovitaminosis, mineral excess / deficiencies)
4. Hormonal (Maternal diabetes, use of synthetic Progesterone, Oestrogen, Cortisone)
5. Maternal infection-viruses (Toxoplasmosis, Syphilis).

Ultrasound scan is currently considered to be a secure, non invasive and cost effective investigation of the foetus. It has progressively become a crucial tool and place an important role within the antenatal care of each pregnant woman. Since its introduction within the late 1950's Ultrasonography has become very useful in Obstetric diagnosis. Currently used equipments are referred to as Real Time Scanners with which endless picture of moving foetus are often depicted on a moving screen. Very high frequency of sound waves of between 3.5 to 7.0 megahertz (3.5 to7 million cycles per second) are generally used for this purpose. They're emitted from a transducer which is placed in touch with maternal abdomen and is moved to seem at any particular content of the Uterus. Repetitive arrays of Ultrasound beams scan the foetus in thin slices and are reflected back to an equivalent transducer. The knowledge obtained from different reflections are recomposed back to an image are often assessed and measurements are often made to each accurate levels on the pictures displayed on the screen. Such measurements form the corner stone within the assessment of fetal age ,size and growth of the foetus.

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Ultrasonography are often performed and repeated without risk to mother / foetus. A scientific anatomical survey of the top, Face, Brain, Spinalcord, Heart, Chest, Abdomen and its contents, Tract , Skeleton and Limbs are administered . Many Structural abnormalities within the foetus are often reliably diagnosed by an Ultrasound scanned these can usually made before twenty weeks. Common examples include Hydrocephalus, Anencephaly, Meningomyelocele, Achondroplasia and other Dwarfism Spinabifida, Exomphalos, Duodenal atresia and Foetalhydrops. With newer equipment, conditions like Cleft lips / palate , Congenital cardiac abnormalities and Mongolism are more readily recognized.

Ultrasonography has become indispensable within the diagnosis / exclusion of Placenta praevia and other Placental abnormalities. Ultrasonography is effective in determining the amount of foetuses and their presentations, evidence of growth retardation and foetal anomaly, presence of Placenta praevia and any suggestion of dual to twin transfusion. A Scan is suggested when an abnormality is suspected on clinical grounds. Otherwise first scan is usually performed at around seven weeks to verify pregnancy to exclude ectopic / molar pregnancies to verify cardiac pulsation and measure Crown-rump length for dating. A second scan is performed at eighteen to twenty weeks to seem for congenital malformations, exclude multiple pregnancies and to verify dates and growth. Placental position is additionally determined. A third scan could also be done at around thirty four weeks to guagefoetal size and assess foetal growth. Placental position is verified. The entire number of scan will verify counting on whether a previous scan has detected certain abnormalities that need follow-up assessment.

Amniocentesis is completed to gather materials for biochemical tests, which can end in a foetal loss of 2% to three. It also requires a culture time varying from two days to three weeks. Ultrasound on the opposite hand, gives immediate result and there's practically no harm to the foetus. Detection rate is additionally higher in Ultrasound screening since we will directly visualize the defects. Ultrasound examination is thus descriptive, which estimation of alpha foeto protein can never be.

This study is to seek out out whether Ultrasound scan reports are supported by gross anatomical defects by autopsy of the foetus. It also aims at checking out the other abnormalities which could are missed by the Ultrasound. The etiology and prevalence of Congenital anomalies have also been studied.

Materials and methods

SOURCE

50 Ultrasound scan reports of pregnant women carrying foetuses with congenital anomalies.Gross study of the specimen of medical termination of pregnancy, which were followed up from the above Ultrasound Scan reports.

Materials

The present study was administered during the year of January 2014 to July 2016. Ultrasound scanning reports of pregnant ladies carrying foetuses with congenital anomalies obtained from Ittyavirah Scan Centre, Thiruvananthapuram. A complete of fifty cases were included within the study.

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Inclusion criteria

Only those Congenitally abnormal foetuses which will be detected by Autopsy, with their Ultrasound Scan reports are available are included during this study.

Exclusion criteria

Foetuses with congenial anomaly where Ultrasound scan reports aren't available aren't included during this study. Each of those cases were followed up and the women were contacted. Case histories were obtained by direct questioning of the patients. A detail history was taken and recorded during a Proforma, giving stress to the subsequent points :

1. Maternal age
2. Period of Gestation
3. History of Consanguineous marriage
4. Socio-economic status
5. History of the drugs taken by the mother during pregnancy
6. History of nutritional deficiency during pregnancy due to starvation, hyperemesis.
7. Exposure to radiation, chemicals, paint.
8. History of abortions / still births.
9. Previous history of Congenital anomalies
10. Family history of Congenital anomalies

The cases were followed up until delivery / until the foetuses are expelled at various hospitals in Thiruvananthapuram. The foetuses after expulsion were also collected the maximum amount as possible. The external appearances of the live babies were carefully examined and noted down for comparison with the Ultrasound reports.

Fifty foetuses were obtained starting from sixteen to twenty weeks of gestation, the fetuses were collected from various hospitals in Thiruvananthapuram. Soon after obtaining, each foetus was placed in 10% formalin, which was the fixative solution used. The foetuses were transported to the Department of Anatomy. The external appearance of every foetus was carefully examined and compared with the Ultrasound report. Then each foetus was dissected out using Scalpel, Scissors and Forceps to seem for internal abnormalities. The other abnormalities which weren't mentioned within the Ultrasound reports were also looked for.

Consent for autopsy was obtained from either of the parent after explaining the necessity. Each foetus was examined consistent with predetermined protocol including Ultrasound diagnosis, Photograph, External and Internal examination. The autopsy protocol included the removal of Thoracic, Cervical, Abdominal and Pelvic organs in block and subsequently dissected into organ block. The Placenta, Foetal membranes and Umbilical cord were studied altogether the cases.

OBSERVATION AND RESULTS

In this study we did a correlation of Prenatal ultrasound with foetal autopsy in fifty MTP specimens of varied Congenital anomalies and classified each anomalies system wise. These anomalies were also classified under four categories.

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Category A- full agreement between Ultrasound and Autopsy

Category B- Autopsy confirmed all Ultrasound findings , but provided additional information of Congenital anomalies.

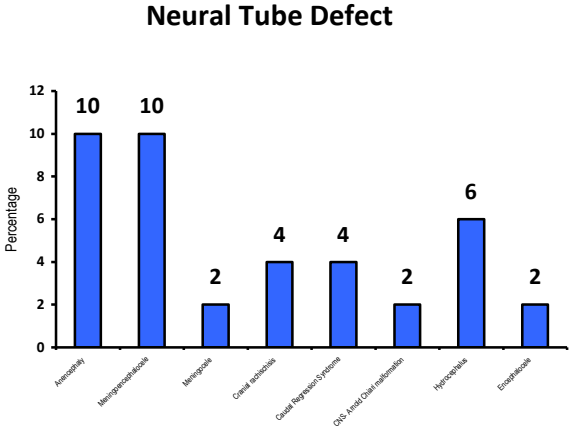
Category C- US findings were only partially demonstrated at Foetal autopsy (some anomalies revealed at US weren't verified
at autopsy).

Category D- total disagreement between US and Autopsy findings. In other words the malformation whose diagnostic
technique led to termination of pregnancy wasn't demonstrated at autopsy.

In Systemwise classification the ectoderm defects comprises the following illustrated in Table 1 and Graph 1 :

Table 1: Central systemanervosum malformations

Neural tube defect	Frequency	Percentage
Anencephaly	5	10.0
Meningoencephalocele	5	10.0
Meningocele	1	2.0
Cranial rachischisis	2	4.0
Caudal Regression Syndrome	2	4.0
CNS-Arnold Chiari malformation	1	2.0
Hydrocephalus	3	6.0
Encephalocele	1	2.0



Graph 1 : Central Nervous System malformations

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Anencephaly

A complete of five cases were obtained. This accounts for 10% of the entire Neural tube defects and 14% of total Congenital anomalies. Five Anencephalic foetuses were obtained, among that four males and one female. All of them had similar features depicted in Figure 1. and the features corresponds with the Ultrasound picture shown in Figure 2.

Anencephaly results from failure of anterior ectoderm closure and occurs before twenty four days of gestation. Anencephaly may be a lethal anomaly characterized by the absence of cerebral hemispheres. Most of the cranial vault is absent. It's the foremost common Central systemanervosum malformation. In neonates, the anomaly is more frequent in females than in males. The danger of recurrence of Anencephaly is 5% to 13%. Anencephaly is related to Myelomeningocele, Microcephaly and Amniotic band syndrome. Extra-cranial abnormalities occur in some cases and included Omphalocele and Clubfoot.

Anencephaly features a multifactorial etiology. Genetic factors seems important due to familial incidence, whereas geographic variation suggests an environmental cause. An increased incidence of Anencephaly and other neural tube.

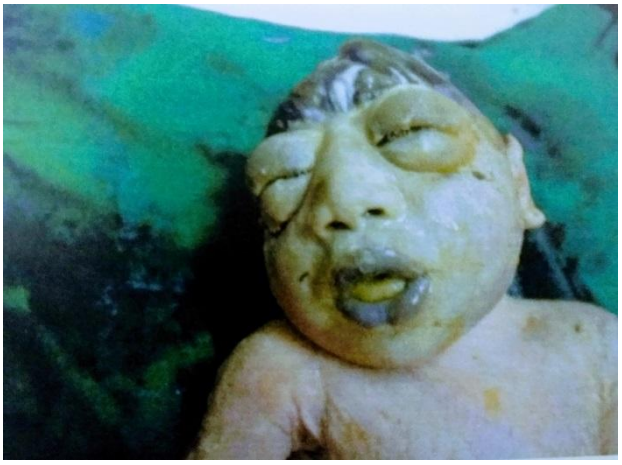

Fig: 01 AnencephalicFoetus

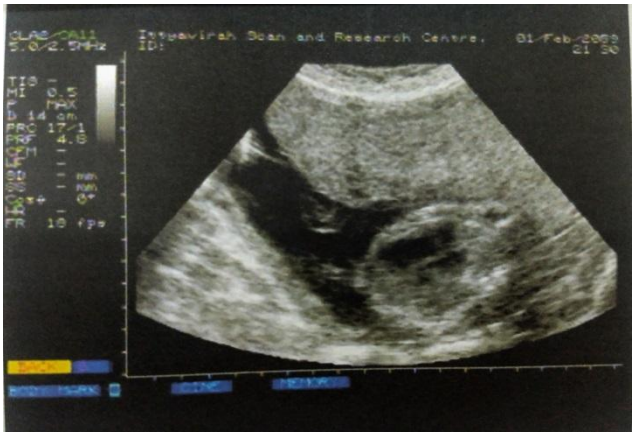

Fig: 02 Ultrasound picture of Anencephaly

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Fig: 03 Foetus with Meningocele

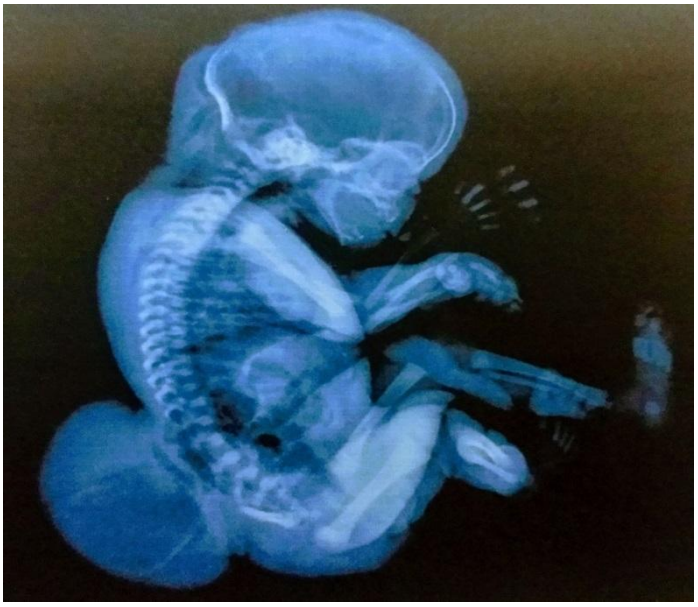

Fig: 04 Xray of Foetus with Meningocele

Meningocele

There was just one case of Meningocele shown in Figure 3. Cerebellum appeared small, Medulla herniated into upper a part of medulla spinalis, Lumbosacral region has Spinal widening and Meningocele, brain slicing showed dilatation of Lateral ventricles depicted in Figure 4.

Hydrocephalus

There have been three Hydrocephalic fetuses. One Hydrocephalic foetus presented a defect within the medulla spinalis below the extent of third vertebra. This is often thanks to the posterior neuropore remaining open. No nerve tissue was seen during this place. Another case of Hydrocephalus was associated with agenesis of Corpus callosum .

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Cystic hygroma

There have been three cases of Cystic hygroma were the pregnancy was terminated within the trimester shown in Figure 5 and Figure 6. One was male foetus with dysmorphicfacies and Occipital meningoencephalocele, along side that ventral Congenital heart defect and Talipes deformity. Second was female baby with generalized Hydrops, Cystic hygroma, serious effusion of all cavities , Bilateral hypoplastic lungs, Preductal tubular hypoplasia of Arch of Aorta. Third one has ambiguous genitalia, hydropic and had ventral congenital heart defect along side Cystic hygroma.

Table 2 : Cystic hygroma

Presence of Cystic hygroma	**Frequency**	**Percentage**
Yes	3	6.0
No	47	94.0
Total	50	100.0

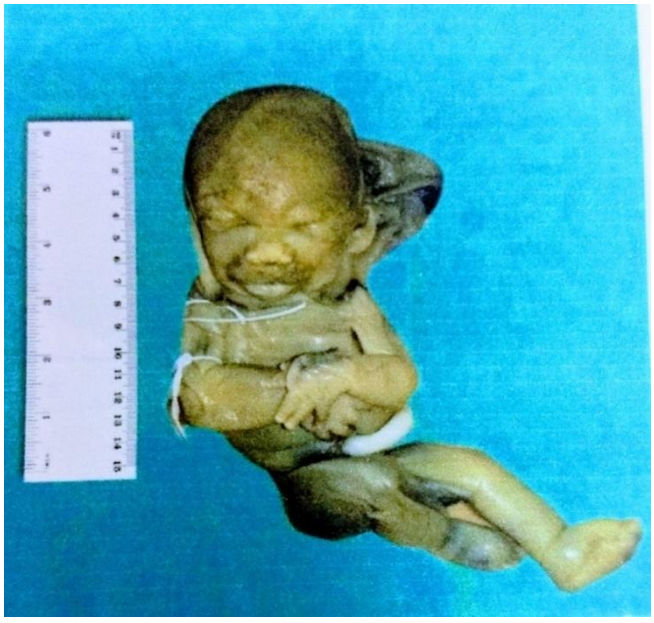

Fig 05: Foetus with Cystic Hygroma

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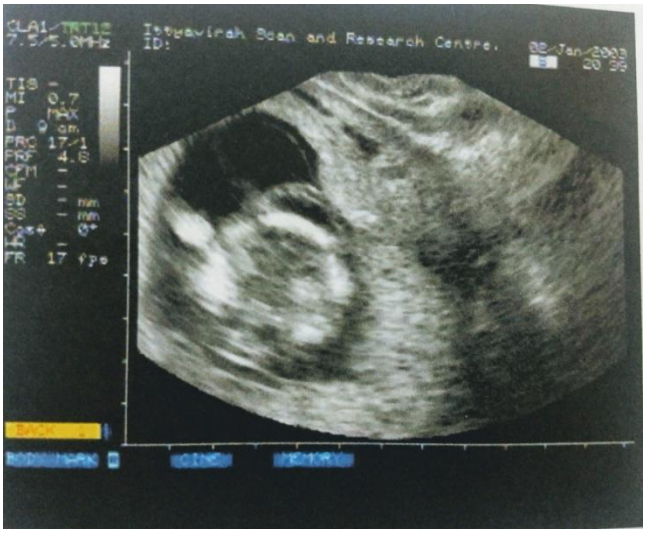
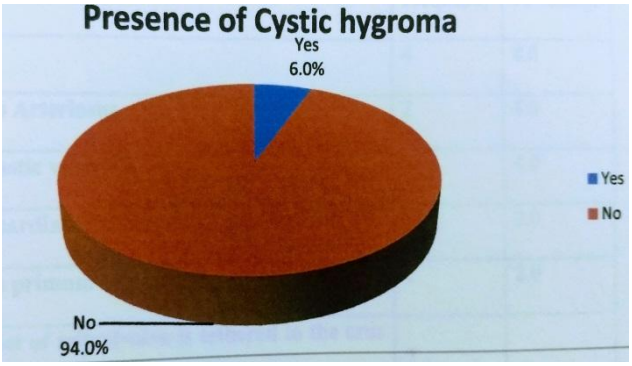


Fig 06: Ultrasound picture of CysticHygroma



Graph 2: Cystic Hygroma

Cardiac Anomalies

Among fifty specimens, fifteen specimens had Cardiac anomalies, four with Ventricular congenital heart defect (8%), one specimen with Dextrocardiasitussolitus, one OstiumPrimum defect with common Atrio Ventricular valve and Anterior leaflet of bicuspid valve is tethered to the crus of Interventricular septum and Endocardial cushion defect each (4%). There have been two specimens of Truncusarteriosus, Hypoplastic ventricle (4%) and one case of Preductal tubular Hypoplasia of Arch of Aorta each (2%) illustrated in Table 3 and Graph 3.

Table 3 : Cardiac malformations

Cardiac anomalies	Frequency	Percentage
VSD	4	8.0
TruncusArteriosus	2	4.0
Hypoplastic Ventricle	2	4.0
Dextrocardiasitussolitus	1	2.0

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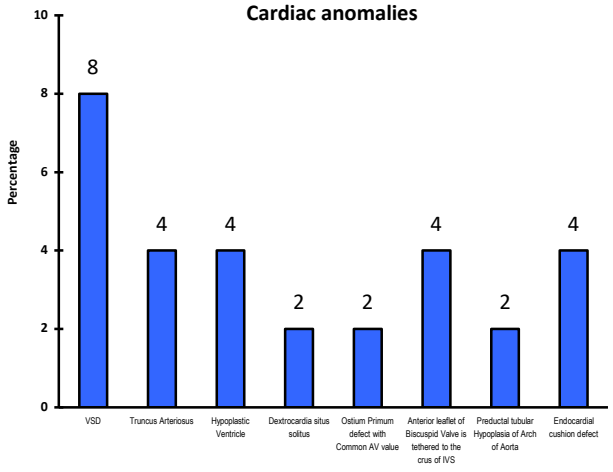
OstiumPrimum defect with Common AV valve	1	2.0
Anterior leaflet of Bicuspid Valve is tethered to the crus of IVS	2	4.0
Preductal tubular Hypoplasia of Arch of Aorta	1	2.0
Endocardial cushion defect	2	4.0



Graph 3: Cardiac Anomalies

Urinary System

Hydronephrosis

Two foetuses of fifty specimens had Hydronephrosis of that one male foetus of approximately twenty weeks size, Kidney shows Hydronephrosis with Glomerular cyst, Bladder dilated thanks to PUV obstruction, Congestion of Internal organs and large retroplacentalhaematoma seen illustrared in Table 4 and Graph 4.

Table 4 : Renal Malformations

Kidney	Frequency	Percentage
Hydronephrosis	5	10.0
Multicystic dysplastic kidney	4	8.0
Polycystic kidney	4	8.0
Cystic kidney with single Ureter	2	4.0
Horseshoe Kidney	2	4.0

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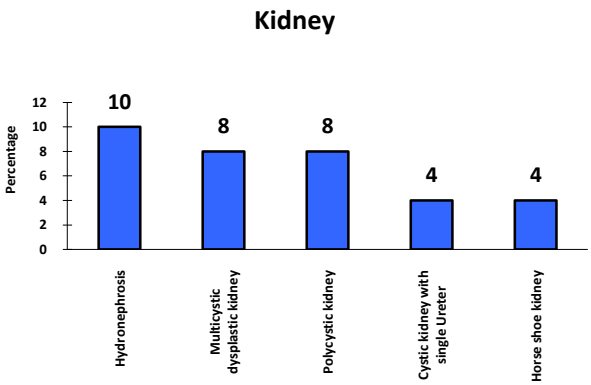


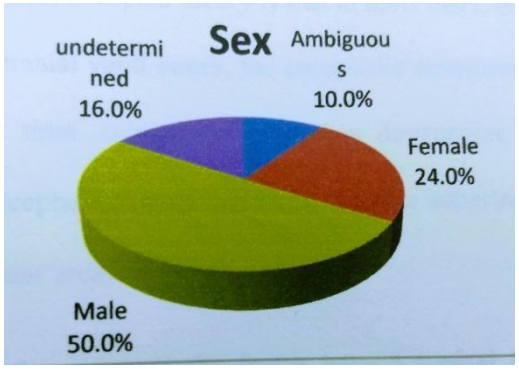
Graph 4: Renal Malformations

Sex Ratio

During this study the frequency of congenital anomalies are more in males (50%), depicted in Table 5 and
Graph 5.which is analogous to the study of RemaDevi et al [(18)]

Table 5: Sex Ratio

Sex	Frequency	Percent
Ambiguous	5	10.0
Female	12	24.0
Male	25	50.0
Undetermined	8	16.0
Total	50	100.0



Graph 5: Sex Ratio ofFoetuses with Congenital Anomalies

DISCUSSION

During this study comparison of Ultrasound which is taken into account as a main method to detect foetal structural anomalies performed till twentieth week of gestation, with Foetal autopsy.

The foremost common Congenital anomalies includes Central systemanervosum defects. In our study we got twenty cases of CNS anomalies which constitute 40% of the entire congenital anomalies. The very best number of CNS malformation we got is Anencephaly, five cases(10%)

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depicted in Table 1. which was accurately diagnosed by Ultrasonography, that correlates with the study of Kaiser et al[26] defects occur in women who have diabetes during pregnancy. Also, women who take Valproic acid for seizure disorder are at increased risk for Anencephaly if their medication has been consumed before conception / during the primary trimester of pregnancy. The foremost widely accepted theory is that in most cases, due to a failure of development of the cranial vault bones, the encephalic structures, covered only by the meninges, are in time subject to extensive destruction, with consequent transformation of the encephalon into a mass of soft tissue adhering to the bottom of the cranium (Cerebral-vascular area).

On Ultrasound, the Anencephalic foetus have a typical frog-like appearance (bulging eyes, harelip / palate, an outsized tongue, and a really short neck) (Chatzipapas and Whitlow,1999). This anomaly is incompatible with life. Approximately 75% of those neonates are still born, and the rest die within the primary hours / days of life. The Anencephalic foetus showed a markedly under developed calvarium, and apart from its basal portions it had been virtually absent. The eyes were large and bulging.

A Sagittal section taken through the top showed a gross deformity of the brain during which only a little nubbin of Midbrain and Brainstem was present. The part of the Skull and the Brain superior to the prominent was practically absent. Forebrain and Midbrain were absent and replaced by rudimentary fibrovascular tissue. Intracranial vessels were underdeveloped. Medulla spinalis was opened, but no abnormality was detected. Both Upper and Lower limbs were normal. Thoracic and Abdominal cavities were opened and looked for the other defects. No abnormality was found.

The second commonest anomalies were of Urogenital system which constitute eighteen cases (36%) of the entire anomalies. This is often almost like the study of Uma Andola[13].We had five cases of Hydronephrosis, illustrated in Table 4 and depicted in Graph 4. Ultrasound revealed dilatation of pelvis, then two cases of Cystic Kidney with Single Ureter, four cases of Polycystic Kidney depicted in Table 4 and illustrated in Graph 4.where Ultrasound showed Cystic dilatation of collecting tubules and bilaterally enlarged echogenic Kidney. Another anomaly during this system include four cases of Multicystic Dysplastic Kidney and two cases of Horse shoe Kidney shown in Figure 7 and depicted in Table 4. The finding is additionally supported by study conducted by Sankar and Phadke[7] where tract malformations constituted the second commonest group of anomalies.

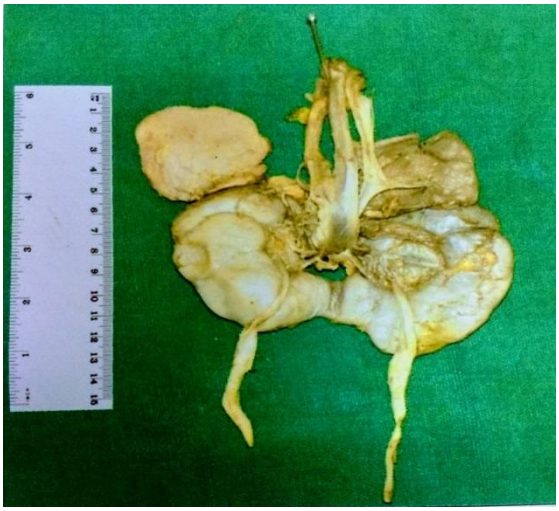

Fig 07: Autopsy Specimen of Horse Shoe Kidney

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The following Multiple Congenital Anomalies were observed :

Two of the cases showed Meckel Gruber Syndrome which incorporates a triad of Central systemanervosum malformation (Occipital encephalocele), Polydactyly (postaxial) and Renal Cystic Dysplasia. It's a lethal, rare, autosomal recessive disorder. It had been first described by a German anatomist ZohannFreebuichMeckel in 1822. Defective gene is present in chromosome 11, 13 and 17.

One case of Arnold Chiari malformation was encountered during which Cerebellum is little, Medulla herniated into the vertebral canal, Lumbosacral canal has spinal widening (Spina bifida) and Meningocele. It had been a female baby with dilated Lateral ventricles and TalipesEquinoVarus both limbs.

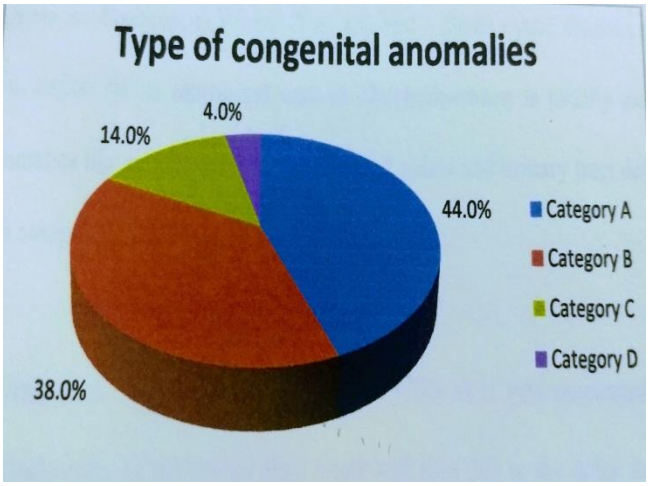
All cases were classified according to the Organ System
(Central nervous system, Cardiovascular system, Gastro Intestinal System, Urogenital apparatus and other multiple anomalies). Finally of those cases were classified into four categories shown in Graph 6. to review the correlation between Ultrasound and Autopsy findings.

Category A- full agreement between Ultrasound and Autopsy

Category B - Autopsy confirmed all Ultrasound findings, but provided additional information of Congenital anomalies.

Category C- Ultrasound findings were only partially demonstrated at Foetal autopsy (some anomalies revealed at Ultrasound
weren't verified at Autopsy).

Category D - total disagreement between Ultrasound and Autopsy findings. In other words, the malformation whose
diagnostic technique led to termination of pregnancy wasn't demonstrated at Autopsy.



Graph 6: Classification of Congenital Anomalies

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Kaiser et al[26] has studied the correlation between Ultrasound and Autopsy in fetuses of all gestational week and evaluated the sensitivity and specificity of Ultrasound towards Autopsy, which doesn't seem to be elevated (40%). Such an evaluation isn't performed during this study. Kaasen et al conducted an equivalent study with foetuses having congenital anomalies, revealed within the trimester Ultrasound scan and terminated[9].

Antonsson et al[6] examined Ultrasound and Autopsy on an equivalent methodological basis and came to a conclusion that autopsy could have significant limitation in CNS malformations. During this study we could ready to notice a high degree of correlation between Ultrasound and Autopsy in twenty two cases. 44% of Foetal anomalies revealed in Ultrasound were demonstrated in Autopsy also. These anomalies are grouped under Category A shown in Graph 6 and depicted in Table 6. which incorporates Ectoderm defects, Cystic hygroma, Hydropsfoetalis and Omphalocele.

Akgun et al[10] in 2007 concluded that analysis of Foetal autopsy following termination of pregnancy enables the diagnosis of anomalies which isn't detected by Ultrasound.

In Category B, limb anomalies, Skeletal defects and Genitourinary anomalies are included mostly. Skeletal dysplasia are often revealed through Ultrasound, but confirmation is required from autopsy. Renal Cystic diseases could even be difficult to define on an Ultrasound scan as Oligohydramnios is usually associated. Other anomalies like Cardiovascular, Cleft lip / Cleft palate and Urinary tract defects are detected in autopsy (Category B) 38% shown in Graph 6 and depicted in Table 6.

Category C included mainly complex, CNS because it gets macerated thanks to improper preservation of the foetus after death and also due to the delay in autopsy. Moreover it should be borne in mind that certain conditions of expulsion hinder examination as they involve an excessively long period of foetal retention resulting in maceration in Utero and tissue lysis, of brain tissue especially and Cardiovascular anomalies like Valve insufficiencies thanks to the tiny size of the guts, Pericardial and Pleural effusions. Seven cases were included in Category C depicted in Table 6. which constitute 14% shown in Table 6 and Graph 6. of the entire anomalies.

In Category D there was total disagreement between Ultrasound and Autopsy. Two cases comes under Category D which constitute 4% depicted in Table 6 and shown in Graph 6. of the entire anomalies.

Table 6: Classification of Congenital Anomalies

Type of Congenital Anomalies	Frequency	Percent
Category A	22	44.0
Category B	19	38.0
Category C	7	14.0
Category D	2	4.0
Total	50	100

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Finally this categorization evidently shows that both Ultrasound and Autopsy may have some important limitations in diagnosing Foetal abnormalities and both are complementary.

The mothers who had undergone Ultrasound scanning and were found to be having Congenitally abnormal foetuses were grouped consistent with the Socio-economic status into High, Middle and Low income groups illustrated in Table 7. This table shows that Congenital anomalies are practically more in Low Socio economic group.

Table 7: Classification consistent with Socioeconomic status

Income Group	Number of Patients
High Income Group	6
Middle Income Group	18
Low Income Group	26

During this study the mothers of fifty foetuses were grouped into three categories age wise illustrated in Table 8. because it was previously believed that the speed of Congenital anomalies increases as maternal age increases.

Table 8 : Classification consistent with the mothers age

Age Group	Below 20 years	21- 30 years	Above 30 years
Number	4	31	15

The table shows that more anomalies were detected in mothers of age bracket 21-30. This result's similar with the study of Mandeep Singh Bindra.[(15)]

In present study it had been found that the Congenital anomalies were high in male babies which is analogous to the study of Kanan et al[(29)] were the ratio of malformed male to female babies was found to be 0.58:0.41. Mohanty et al[(30)] also reported higher incidence of congenital malformations in male babies (1.91%) than in female babies (1.27%). Aiyar and Agarwal[(34)] also observed that the very best incidence of malformation was among term normal weight babies and have documented male preponderance amongst Congenital malformed babies.

CONCLUSION

This study reflects the potential and limitations of both Ultrasound and Autopsy. Due to this limitations of both the techniques, categorization of this study into four (Category A,B,C,D) shown in Graph 6 and Table 6.was possible. Even though Ultrasound reasonably detects the malformations ,Foetal autopsy is important for getting additional information.

In couples having babies with birth defect, diagnosed by Ultrasound and confirmed by Autopsy could also be given counseling regarding the prospect of getting similar anomalies in subsequent pregnancies. These guidance given to the oldsters should be unbiased and respectful to the patients choice, culture, religion and beliefs. Couples should be told about the Ultrasound findings which is confirmed by findings in Autopsy. When a pregnancy is terminated due to anomalies detected by Ultrasound scan and if the autopsy is declined, parents will remain ignorant about the danger of recurrence in subsequent pregnancy.

Ultrasound findings depend upon the accuracy of the machine and the expertise of the person which is like the skill of Autopsy expert in interpreting the anomalies.

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In cases like Category C, where Ultrasound provides information which is difficult to verify with the assistance of Autopsy. The explanations for this disagreement might be the fault in preserving foetal brain which results in the failure in detecting Central systemanervosum malformations during Autopsy. Foetal brain get macerated very soon which fails to offer relevant autopsy report regarding Central systemanervosum malformation of an equivalent foetus. So after the medical termination of pregnancy, the specimen should be frozen as early as possible for attaining a top quality autopsy report. Regarding the cardiac anomalies detected in Ultrasound may remain hidden in Autopsy due to the tiny size of the Foetal heart. Additionally Ultrasound are often considered as a dynamic examination to reveal the functional changes which is difficult to detect in autopsy like flow inversion in aorta and valve insufficiencies.

In certain cases Ultrasound has its own limitations like, detecting the Central nervous system malformations thanks to some factors a bit like the non-specific appearance of some anomalies, the technical factors which can complicate visualization of the brain near the transducer and of the posterior fossa (especially late in gestation), and the parenchymal abnormalities that often cannot be visualized (like Schizencephaly). Moreover Ultrasound evaluation of the spine could be limited by Oligohydramnios. Maternal Obesity and the position of the foetus. Shadows formed from the bony structures can also preclude a whole evaluation of the spinal cord and Arachnoid sac. Also demonstration of tract on Ultrasound is difficult. So Ultrasound can miss variety of those diagnosis. Wagn et al suggested that thanks to these limitations plenty of studies have compared Resonance Imaging and Ultrasound for diagnosis of Central nervous system anomalies, remarking that Resonance Imaging are often an honest supplement to Ultrasound in complicated pregnancies.

Placental examination is additionally useful in verifying infection and in cases where maternal factors play an outsized role in pregnancy outcome. Fresh examination of placenta is preferred for better assessment of weight and discolouration because it increases its weight from 6% to 10% after formalin fixation. Macroscopic fixation of placenta are often done portion Umbilical cord, Disc proper (Foetal surface, Maternal surface and Cut surface) and Extraplacental membrane.

Nowadays when Ultrasound detects a foetal anomaly were medical termination is indicated, that there has been a rise within the rate of termination of pregnancy, but there has been a decline within the autopsy rate. When a diagnostic procedure was supported the results of a scan only, the addition of data from an Autopsy by a Specialist Paediatric Pathologist is missing, which can be a crucial information which will change the estimated risk of recurrence.

In developed countries Congenital malformations are the foremost dominant explanation for infant morbidity and mortality[17].Treatment and Rehabilitation of those morbid children is difficult and recovery is typically impossible. Here only the importance of early recognition of anomalies is known .

The foremost of Congenital anomalies are of Multifactorial causation like Genetic factors (Chromosomes, Single gene mutations), Environmental exposures, Infectious agents, Chemical compounds, Radiation, Use of medication, Maternal metabolic diseases, Multiple births, Maternal life event stress, Prematurity and Occupational exposure. Furthermore, Low Socioeconomic status is additionally an element which are highly relevant. Environmental exposure are often a Preconceptional mutagenic action / a Postconceptionalteratogenic action. Deficiency of Folic acid within the Preconceptional period are established risk factor for ectoderm defects. Various studies show that incidence of anomalies is increasing in elderly

pregnancies and in pregnancies which aren't monitored. Consanguineous marriages are also one among the important factors contributing to the increased rate of Congenital malformations, gene which was hidden for generations may thus come to light for the primary time. Due to high consanguinity rates within the Muslim population, the incidence of Congenital anomalies in Islamic countries is between 10 to 45%.[17]

Maternal age is a crucial parameter within the birth of a Congenitally malformed foetus. For this reason, females who are older than 35years aged got to be examined more carefully since the danger of birth of a congenitally malformed foetus increases.

Foetal autopsy is emerging as a useful tool for diagnosis of foetal abnormalities, many of which may go undiagnosed by Ultrasound evaluation. Consulting Obstetrician do understand foetal abnormalities better most of which may be diagnosed by Sonography and can therefore optimize the counseling process and plan Prenatal / Postnatal intervention. A better evaluation by Autopsy helps in counseling the patient with reference to subsequent pregnancies. It's hoped that a correct Postmortem evaluation of foetus will help in better patient counseling and plan subsequent pregnancies. Although these results are encouraging, there's still an extended thanks to go.

In couples with history of Congenitally anomalous children, with the assistance of recent investigative procedures like Antenatal Sonography, Counseling and proper planning for subsequent pregnancies might be an eye fixed opener for a healthy future generation.

Conflicts of interest

All authors have none to declare.

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