DEPARTMENT OF OBSTETRICS & GYNAECOLOGY

MADURAI MEDICAL COLLEGE

MADURAI

CERTIFICATE

This is to certify that, this dissertation titled **"STUDY OF MIDTRIMESTER ANOMALY SCAN"** submitted by **Dr.M.VENNILA** to The Tamilnadu Dr.M.G.R. Medical University, Chennai in partial fulfillment of the requirement for the award of M.S. in (OBSTCTRICS AND GYNAECOLOGY) branch III is a Bonafide research work carried out by her under our direct supervision and guidance.

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DECLARATION

I, Dr.M.VENNILA, hereby declare that, I carried out this work on "PROSPECTIVE STUDY ONMIDTRIMTER ANOMALY SCAN AND ANALYSIS in **GOVT.RAJAJI HOSPITAL**" at the Department of **OBSTETRICS & GYNAECOLOGY**, Govt.Rajaji Hospital, Madurai, under the guidance of **Prof.Dr.K.S.CHITHRA** Professor of OBSTETRICS & GYNAECOLOGY, during the period of JUNE 2013 to JULY 2014. I also declared that this bonafide work has not been submitted in part or full by me or and others for any award, degree or diploma to any other University or Board either in India or abroad.

This is submitted to The Tamil Nadu Dr. M.G.R. Medical University, Chennai in partial fulfillment of the rules and regulations for the M.S.degree examination in Obstetrics & Gynaecology (Branch II) to be held in April 2015.

Place : Madurai

Date : **(Dr.M.VENNILA)**

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Ref. No. 23308/E4/2/2013 **Govt. Rajaji Hospital, Madurai.20. Dated: 24.12.2013**

Institutional Review Board / Independent Ethics Committee.
Dr. N. Mohan, M.S., F.I.C.S., F.A.I.S.,
Dean, Madurai Medical College &
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Sub: Establishment-Govt. Rajaji Hospital, Madurai-20-
Ethics committee-Meeting Minutes- for December 2013
Approved list -regarding.

The Ethics Committee meeting of the Govt. Rajaji Hospital, Madurai was held on 18.12.2013, Wednesday at 10.00 am to 12.00.noon at the Anaethesia Seminar Hall, Govt. Rajaji Hospital, Madurai. The following members of the committee have attended the meeting.

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The following Project was approved by the committee

..2..

Name of P.G.	Course	Name of the Project	Remarks
Dr. M. Vennila	P.G in M.S., (O&G), Madurai Medical College and Government Rajaji Hospital, Madurai.	Prospective study on detection of congenital anomalies by targeted scan at 18 to 20 weeks of gestation	Approved

Please note that the investigator should adhere the following: She/He should get a detailed informed consent from the patients/participants and maintain it Confidentially.

1. She/He should carry out the work without detrimental to regular activities as well as without extra expenditure to the institution or to Government.
2. She/He should inform the institution Ethical Committee, in case of any change of study procedure, site and investigation or guide.
3. She/He should not deviate the area of the work for which applied for Ethical clearance. She/He should inform the IEC immediately, in case of any adverse events or Serious adverse reactions.
4. She/He should abide to the rules and regulations of the institution.
5. She/He should complete the work within the specific period and if any Extension of time is required He/She should apply for permission again and do the work.
6. She/He should submit the summary of the work to the Ethical Committee on Completion of the work.
7. She/He should not claim any funds from the institution while doing the work or on completion.
8. She/He should understand that the members of IEC have the right to monitor the work with prior intimation.

Member Secretary **Chairman**
Ethical Committee

DEAN/Convenor
Govt. Rajaji Hospital,
Madurai- 20.

04/12/13

To
The above Applicant
-thro. Head of the Department concerned

“STUDY OF MIDTRIMESTER ANOMALY SCAN”

CONTENTS

INTRODUCTION

INTRODUCTION

Prenatal diagnosis is the art and science of identifying structural functional abnormalities in developing fetus.2-3% of all neonates have major abnormalities discovered at birth.

Ultrasonography is widely used for the prenatal evaluation of growth and anatomy of the fetus. The main objective of a routine mid-trimester fetal ultrasound scan is to provide accurate diagnostic information for optimized antenatal care with the best possible outcomes for mother and fetus. Many fetuses with major chromosomal abnormalities have isolated or multiple structural abnormalities which can be recognized on 18 to 20 weeks anomaly scan.

Prenatal diagnosis of anatomic anomalies is very important in making decision about therapeutic termination of pregnancy.

Ultrasound detection of prenatal abnormalities is a safe, readily available noninvasive technique which is more acceptable by patients. Several studies have shown an accuracy of more than 95% for ultrasound detection of prenatal anomalies, with no confirmed biological hazards.

Care of handicapped and disabled person is a serious health care burden on communities as well as it affects the parents. Antenatal diagnosis of congenital anomalies is of great help to health economy.

It is recommended by the Royal college of Obstetrics and Gynaecology that all women should be offered a fetal anomaly scan at 18 to 20 weeks of gestation (RCOG 2007).

AIM OF STUDY

1. Identify serious fetal abnormalities either incompatible with life or associated with morbidity.
2. Identify certain abnormalities that require early intervention following delivery
3. Offer choices to women about their further screening options and pregnancy management.
4. To find any causal association between anomalies and consanguineous marriage or nutritional deficiencies or drug intake or environmental influences
5. To counsel the women about how to avoid anomalies in forthcoming pregnancies. eg.strict control of diabetes and periconceptional folic acid intake in neural tube defects.

REVIEW OF LITERATURE

1. Ian Donald used sonar extensively in obstetric practice. He did stage I sonar study to diagnose early pregnancy complications, fetal maturity, placental localization and diagnosis of gross anomalies.
2. Stuart Campbell (1983) used sonar in detecting anomalies and reported the incidence of false positives and false negatives.
3. Johnson et al (1983). Stork et al (1985) have done work on diagnosing fetal anomalies with Nuclear Magnetic Resonance. They claim that still USG is ideal because real time technique is not present in magnetic resonance imaging.
4. Hobbins et al (1980) reported prenatal diagnosis of hydrocephalus by sonar in II trimester.
5. Saari - Kemppainen and Colleagnes (1990) conducted a randomized study of routine ultrasound between 16 -20 wks and showed that perinatal mortality was significantly lower in sonography group largely because of pregnancy termination for malformed fetus.
6. Luck (1992) did routine USG at 19 weeks of gestational age and concluded that this approach reduces perinatal morbidity and mortality because of termination of crippling or lethal malformations.

7. Sabbagha (1987) in his study has reported major anomalies in approximately 5% of live born infants and minor anomalies in 15% of all infants.
8. Rajan (1993) has reported that in obstetric ultrasound studies major fetal malformations have been detected, giving incidence of 3% common anomalies included CNS malformation - 31.82%, renal tract anomalies 16.98% ventral wall defects 15.10%. Cardiac 9.4% Skeletal 9.4% and GIT obstructions 4.70%.
9. RADIUS TRIAL: Routine Antenatal Diagnostic Imaging with Ultrasound trial was sponsored by National Institute of child Health and Human Development and results reported by Ewigman and Colleagues (1993) concluded that routine screening for low risk women did not improve perinatal outcome.

HUMAN EMBRYOLOGY

There are 3 basic stages in human development. The First stage begins with fertilization and lasts until organ formation begins at the start of 3rd week. The zygote is the fertilized egg prior to the I^{st} cell division. The embryonic stage begins with the first cell division and lasts until the end of 8^{th} week. By this time all organ have begun to form or have completed their formation. Organogenesis from 3^{rd} to 8th week is second basic stage. The final fetal stage is primarily one of growth and maturation of previously formed structures.

Earlier, most birth defects were thought to arise during organogenesis and were only malformations. Now it has been found that many anomalies are not malformation and those congenital anomalies many develop at any tissue from the beginning of embryogenesis to term.

The first two weeks period of ovum phase is not associated with birth defects. An insult which occurs during this period either destroys the embryo completely or leaves it unaffected. Earliest defect arises at the embryonic disc stage (ie from 2nd week onwards) At this stage 3 basic tissue types- the ectoderm, endoderm and mesoderm are forming and any malformation at this time occur as a result of aberrant formation of mesoderm secondary to abnormal migration of mesodermal precursor cells.

HUMAN EMBRYOLOGY – DEVELOPMENT AT EARLY WEEKS OF GESTATION

Days of embryonic life (days)	Crown rump length (M.M)	Ext. features – skeletal system and general body	CNS and sensory organ development	Cardiovascular and respiratory system development	Digestive system	Genito urinary system
0	0	Ovum, zygote Blastocyst implantation	-	-	-	-
10	1	-	Neural plate	Cardiogenic plate	Fore and hindgut	-
20	2	Somites Anterior and Posterior limb buds	Optic vesicles, otocyst anterior neuropore closes posterior neuropore closes.	Heart tubes, Ist Aortic arch Lung primorida, septum primum, aortic arches	Liver bud, stomach primodrium, dorsal pancreas	Pronephros, mesonephros
30	4	Myotomes	Olfactory pits	Primary and labor bronchi, septum secondum	Ventral pancreas, spleen, umbilical hernia, division of cloaca	Metanephros, genital ridge

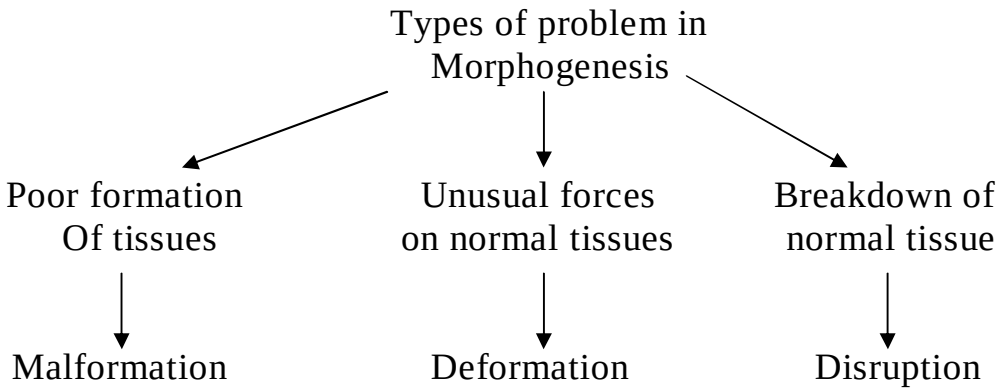
40	10	Finger rays, cartilagenous	Primordium of cerebellum, retinal pigment, pinnae formation	Ductus venosus division of truncus	-	Primodium of gonad
50	20	Primary ossification centre, separate digits. Palatal process grow medially	Eyelids, semi circular canal, differentitation of cerebral cortex	Septa complete, segmental bronchi	Rectum and Bladder separated lumen in gall bladder	Gonadal differentiation mullerian primordium
60	30	Palatal fusion	Almost CNS completely developed	Main blood vessels present	Umbilical hernia reduced	Uterus complete gonad differentiated

ETIOLOGY OF BIRTH

DEFECTS

ETIOLOGY OF BIRTH DEFECTS

Birth defects can occur in at least 3 ways.



1. ***In malformation,*** the fetus or structure is genetically abnormal and thus programmed to develop abnormally. Malformation occurs from beginning of organogenesis to l2th week of post conceptual development. eg limb contracture resulting from diastrophic dysplasia.
2. ***Deformation*** is when a genetically normal fetus develops in an abnormal uterine environment causing structural changes. They can occur at any time from embryonic disc stage onwards, but more common in late half of III trimester when fetal crowding can occur, eg is oligohydramnios causing limb contractures.

3. ***Disruption*** occur when a genetically normal fetus suffers an insult resulting in disruption of normal development . This can occur at any time, eg amnion rupture causing limb deformities.
4. ***Dysplasia*** Abnormal organization of cell or extra cellular substance within tissue, which may result in alteration in tissue morphology.
5. ***Phenocopies*** These are identical appearing abnormalities but can have widely varying etiologies. Multiple Structural defects or developmental abnormalities can also occur together in one individual.
6. ***Syndrome*** It is a cluster of several anomalies or defects which are embryologically unrelated and occur together at relatively high frequency that have the same cause (eg) Trisomy 18.
7. ***Sequence*** When single defect leads to development of one or more abnormalities eg oligohydramnios leading to pulmonary hypoplasia. limb contractures and facial deformities.
8. ***Association*** These particular anomalies occur together frequently, but do not seem to be linked etiologically (eg) CHARGE association which is a combination of coloboma, heart defects, atresia choanae, mental retardation, growth deficiency and ear anomalies. VATER association – Vertebral anomalies, imperforate anus, tracheoesophageal fistula with oesophageal atresia and renal, radial and cardiac defects together.

Categorization of congenital anomalies (Brent, 1979)

Genetic	Genetic mutations Chromosomal Aberrations	20% 5%
Environmental	Fetal infections (mainly viruses) Maternal diseases (endocrine and metabolic abnormalities etc) Mechanical factors Drugs and Chemicals	3% 3-4% 1-2% 1%
Multi factorial		65%

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Incidence of congenital anomalies may be influenced by maternal age, parity, race, consanguinity, sex of the infant and environment.

Excessive aging of the egg prior to fertilization seems to be a cause for some other malformations. With the increased awareness of factors influencing malformation, when the mother is exposed to such factors, it is important to rule out malformation in growing fetus.

1. Chromosomal abnormalities

Chromosomal abnormalities are the cause for about 50% embryonic deaths. 5.7% total losses, 6-11% still births and neonatal deaths and 0.91% live born children.

(i) Aneuploidy

There are total 44 autosomes arranged in pairs numbered 1-22 and I pair of sex chromosomes. Most obvious or early recognized chromosomal abnormality are numerical. If affected individual inherits an extra chromosome— trisomy. missing a chromosome - monosomy or has an abnormal number of haploid chromosome complement - polyploidy. Incidence of chromosomal abnormality is 9 per 1000 derived from data using high —resolution band (350-550 band) which allows accurate identification of deletions and rearrangements. Most trisomies and nonmosaic monosomies result from abnormality in meiosis.

(ii) Chromosomal deletions

Deletion refers to a portion of chromosome that is missing. Most deletion occur during meiosis and result from malalignment or mismatching during pairing of homologous chromosomes.

Eg. Del 5p is also called Cri du chat syndrome. If deletion is -identified parents should be tested to find whether it is associated with a familial translocation that would have a recurrence risk.

(iii) Chromosomal translocation - Reciprocal, , Robertsonian, Isochromosomes

Reciprocal translocation Rearrangement of chromosomal material in which breaks occur in 2 different chromosomes. The fragments are exchanged before breaks are repaired. Rearranged chromosome is called derivative (der) chromosome. If no chromosomal material is gained or lost in this process, it is a balanced translocation. Carriers of a balanced translocation can produce unbalanced gametes that result in abnormal off spring.

Robertsonian Translocation Result from centric fusion of 2 acrocentric chromosomes which are 13,14,15,21 and 22. These are common with incidence of 1 in 1000.

Isochromosomes Composed by either 2q (long) arms or 2p (short) arms of one chromosome fused together. There are thought to arise when the centromere breaks transversely instead of longitudinally during meiosis II or mitosis. The other chromosomal abnormalities are chromosomal inversion, ring chromosomes and chromosomal mosaicism.

2. Single Gene Defects

Single gene defects are sometimes responsible for gross anatomical malformations (eg achondroplasia) but more commonly cause abnormalities at molecular level. They may manifest in heterozygote or homozygote. Although

transmission pattern of these disease are consistent with Mendelian inheritance their phenotypes are strongly influenced by modifying genes and environmental factors.

3. Hormonal Preparations

Progestational agents have been used for treating recurrent abortion. Ethisterone or nor-ethisterone is capable of causing virlization in female fetuses - clitoral hypertrophy or labial fusion.

Mothers exposed to DES or any estrogenic supplementation during Ist trimester may have female offsprings with vaginal, Cervical abnorma1ities in 30%.

Cortisone cause cleft palate and lip. Antithyroid drugs including iodine may depress fetal thyroid function, leading to congenital hypothyroidism. Goiter may be observed in few cases. 2.3% of all congenital malformation can be attributed to the drugs or other chemicals in the environment.

4. Infections

Viruses and other infective agents can affect the growth and development of fetus in 2 different ways. Infection during the period of organogenesis results in malformations. But in late pregnancies it causes growth retardation of the fetus. These two manifestation sometimes overlap.

i. Rubella virus – Cataract, Micro ophthalmia, Deafness, CHD

The discovery of association between Rubella (German measles) in pregnancy and congenital cataracts in offspring was made by late Sir Norman Gregg in 1941.

ii. Cytomegalovirus **90**% of cases congenitally infected with this virus is asymptomatic while 10% have fetuses with CNS defects. Damage done by. this virus in not amenable to correction. It's difficult to diagnose this viral infection.

iii. Toxoplasmosis Toxoplasmosis Gondii is a protozoan. This is associated with placental disease. Intrauterine infection results in abortion, still birth and congenital neurological damage.

iv. Herpes Simplex Diffuse brain damage, mental retardation. Microcephaly intracranial calcification micro- ophthalmia. Retinal dysplasia and chorioretinitis are some of the sequelae of Herpes simplex viral infection.

v. Others

Chicken pox	-	Muscular atrophy, mental retardation
Venezuelar equine Encephalitis	-	Cerebral defects, cataract
Syphilis	-	Dental anomalies. skeletal anomalies, mental retardation.

5. Chemicals and Pollutants

Obstetricians should be aware of the types of pollution due to agriculture and industry. The nitrate pesticides, insecticides, fungicides. rodenticides, herbicides are the common pollutants. This may enter maternal circulation but symptoms may not be present. Dioxin, a byproduct in chemical industry causes cardiac and brain malformations.

6. Occupational Hazards

Pregnant woman should not be allowed to work in occupation involving exposure to harmful chemical substances that may produce systemic damage, anoxia and irritation of the respiratory tract. Medical lab workers are also exposed to many infective agents. Incidence of birth defects in offspring of laboratory workers are 2-3 folds higher than normal.

Its observed that there is an increased risk of congenital anomalies, stillbirth, and abortions among women anaesthetists and theatre nurses.

7. Stress in Pregnancy

The stress in pregnancy is pre-conceptional. It is suggested that endocrine disturbances resulting from emotional stress may he constantly associated with chromosomal anomalies found in Downs syndrome.

8. Mechanical Factors

Skeletal anomalies of fetus are observed due to malformed uterus fetal compression giving rise to talipes, congenital dislocation of hip, arthrogyphosis, Constriction ring due to amniotic band causes congenital amputation, the extent varying from tip of one of the digit to an entire limb.

9. Ionizing Radiation

When the patients are exposed unknowingly to ionized radiation during Ist trimester or when patient requires exposure during pregnancy it causes deleterious effects on growing fetus. Radiation has both cumulative and single dose effect. The degree of damage is dose dependent. The dose varies according to tube velocity (kv) tube current (ma), time of exposure, film distance, size of field and thinness of aluminum fitters Dosage is to be calculated and to be individualized. Amount of irradiation received by fetus during single assay ranges from fraction of a rad to 2-2.5 rads.

Accidental irradiation has caused microcellular defects, blindness and other anomalies. Radiation is a mutation inducer and also a carcinogen apart from teratogenic activity.

10. Nutrition

Folic acid is essential for production of methionine. Which is a co-factor in RNA and DNA synthesis and is required for methylation of protein, lipid and myelin.

In humans, deficiency of folic acid in the preconceptional period is known notoriously to cause neural tube defects. The association between folic acid deficiency and anomalies has been widely studied and folic acid is now recommended in periconceptional period to mothers.

11. Consanguinity

Studies have concluded that consanguinity leading to anomalies are more marked when consanguinity was practiced in more than- one generation.

12. Polyhydramnios and Oligohydramnios

Amniotic fluid is constantly being produced and removed depending on the efficacy of fetal urinary system and swallowing. Classically oligohydramnios is seen in renal agenesis, but it may also occur with obstructive lesions in urinary tract and with polycystic kidneys.

Association of polyhydramnios in anencephaly probably results from hyper extension of the neck interfering mechanically with swallowing and not from any deficiency of swallowing reflexes. Its also due to exposed part of brain largely composed of choroids plexus secreting excessive amount of fluid. 0f more

practical importance is association of polyhydramnios with high intestinal obstruction especially common with esophageal atresia (10-20%) fairy common with duodenal atresia. and relatively rare with ileal and lower bowel obstruction. A tube should he passed into the stomach of an apparently normal baby horn to a mother with polyhydramnios to exclude existence of esophageal atresia.

13. Teratogens

Although congenital malformations caused by drugs and chemicals have attracted much attention, they account for only about 10% of the total and only a small amount of it is identified.

Before labeling any agent as a potent teratogen, certain criteria must he met with

1. Defect must be completely characterized.
2. Agent must cross the placenta
3. Exposure must occur during a critical developmental period.
4. Cause and effect must he biologically plausible.
5. Epidemiological studies must be consistent.
6. Suspected teratogen causes a defect in an animal.

Teratogens probably act by disturbing specific pathogenic processor, leading to cell death, altered tissue growth, abnormal cellular differentiation or disruption of normal development. Some teratogens disrupt one or more these processes and combination of drugs may be additive. Two established mechanism

of teratogenesis are disruption of folic acid metabolism and production of oxidative intermediates.

i. Drugs

Drugs		**Major Abnormalities**
Thalidomide	-	Malformation of all 4 limbs
Diethyl stillbestrol	-	Vaginal cancer
Synthetic corpusluteum hormone	-	Masculinization
Warfarin (anticoag)	-	Nasal hypochondroplasia, Abnormalities of CNS
Hydantoin (anticonvulsunt)	-	Facial abnormities, microcephaly, Mental retardation
Trimethadione (anticonvulsant)	-	Growth retardation, cleft lip, cleft palate.
Aminopterin (folate antagonist)	-	Spontaneous abortion Hydrocephalus
Streptomycin	-	Hearing loss
Tetracycline	-	Dental pigmentation, Enamel Hypoplasia
Valproic Acid	-	Neural tube defects
Vitamin A acid	-	Cranial and facial anomalies, Neural tube defects
Lithium	-	Cardiac malformation
Anti-thyroid drugs	-	Hypothyroidism, thyroid tumors
Isotretinoin	-	Agenesis of the ears, multiple CNS defects, facial and CVS anomalies.

ii. Industrial Chemicals

Lead	-	Infertility, spontaneous abortion, Mental retardation
Mercury	-	Still birth
Methyl mercury	-	Cerebral palsy, microcephaly
PCB	-	Spontaneous abortion, still birth, Pigmentation of skin and teeth
Organic solvents (tolunene)	-	Low birth weight, birth defects
Ethylene glycol	-	Spontaneous abortion, infertility (decrease sperm count), birth defects
Cadmium	-	Infertility, delayed neurological Development
Heat	-	Infertility, decreased sperm count.
Radiation	-	Microcephaly, metal retardation.

iii. Others

Alcoholic beverages	-	Growth retardation, microcephaly, Metal retardation
Tobacco smoking	-	Spontaneous abortion, IUGR
Diabetes Mellitus	-	Congenital heart disease,

		malforamations of lower trunk.
Iodine deficiency	-	Thyroid tumours, mental retardation growth retardation.
Maternal phenylketonuria	-	Microcephaly, mental retardation.

Recently endocrine disruptors have been found to exert a toxic effect mainly by binding to estrogen receptors on cells and thus supporting or enhancing the actions of estrogens. But these have to be studied extensively.

PRENATAL DIAGNOSIS

At the present era, almost many if not all disease and anomalies affecting the fetus can be diagnosed prenatally and according to its severity termination or fetal therapy can be considered.

The various modalities by which prenatal diagnosis can be done are

1. Radiography
2. Ultrasonography
3. Electro cardiograph
4. Estimation of serum α feto protein
5. Examination of amniotic fluid for
    a. Biochemistry of fluid
    b. Cytology of fixed cells
    c. Cytogenesis of cultured cells.
    d. Biochemistry of cultured cells.
6. Sex determination by endocervical smears
7. Fetoscopy
8. Fetography
9. Chorionic villi biopsy
10. Percutaneous umbilical cord blood sampling (PUBS)
11. Fetal tissue biopsy
12. Preimplantation diagonosis
    - Polar body analysis
    - Blastomere biopsy
13. Analysis of fetal cells in maternal circulation

1. Radiography

Fetal radiography is the most helpful in demonstrating defects of the skeletal system. Anencephaly can be readily recognized but spinabifida, hydrocephalus is quite difficult to diagnose. Less common defects including achondroplasia and conjoined twins can be diagnosed before birth.

2. Electrocardiography

Fetal ECG has little part to play in prenatal diagnosis of congenital anomalies but one important condition - congenital heart block can be recognized during or before labour.

3. Serum Alpha feto protein estimation

This glycoprotein is synthesized by fetal yolk sac early in gestation and later by gastrointestinal tract and liver. It circulates in fetal serum and passes into fetal urine and amniotic fluid AFP passes into maternal circulation by diffusion across the placental membranes and may also be transported via placental circulation. It steadily increases in maternal serum after 12 weeks. When there is fetal abnormality such as opening uncovered by integument somewhere in the body or number of other problems, then maternal AFP serum levels are increased.

Maternal serum alpha feto protein is usually done between 15-22 weeks. Factors that influence the result include maternal age, weight, race, diabetic race, number of fetuses as well as gestational age determined by accurate menstrual history or sonography. Maternal serum AFP is measured in ng/ml and reported as a multiple of median (MOM) of the unaffected population. APP $\geq$ 2.0-2.5 MOM indicates increase risk and warrants further evaluation.

The following are examples of conditions with abnormal maternal serum AFP concentrations.

a. Elevated Levels Neural tube defects, pilonidal cyst. eosophageal or intestinal obstruction. liver necrosis, cystic hygroma, sacrococcygeal ieratoma, abdominal wall defects, omphalocele. gastroschisis. urinary obstruction. renal anomalies-polycystic or absent kidneys, congenital nephrosis, osteogenesis imperfecta, congenital skin defects. cloacal extrophy, chorioangioma of placenta, low birth weight, oligohydramnios, multi fetal gestation, decreased maternal weight, Underestimated gestational age, maternal hepatoma or teratoma.

b. Low levels Chromosomal trisomies, gestational trophoblastic disease, fetal death, increased maternal weight and over estimated gestational age.

4. Amniocentesis

Amniocentesis for genetic diagnosis is usually performed between 15-20 weeks. Under USG guidance 20-22 gauge spinal needle is introduced into the

amniotic sac carefully avoiding the placenta, umbilical cord and fetus, and amniotic fluid is taken for analysis. Number of multicenteric studies have confirmed its safety and it has more than 99% diagnostic accuracy.

5. Prenatal sex determination by Endocervical smears

Endocervical smears are stained with 0.5% quinacrine hydrochloride and visualized under fluorescent microscope. Presence of Y fluorescent body in more than 3% of cells in highly suggestive of male fetus. Orcein staining is done to visualize barr bodies in cells presumed to be of fetal in origin. Presence of Barr bodies over 20% indicates female fetus.

6. Fetoscopy

Direct examination of fetus using a modified infant cystoscope was introduced by Valenti (1972) and subsequently developed by Scrimgeour (1974).

7. Fetography

Is extremely useful to detect fetal abnormalities that may be associated with polyhydramnios.

8. Chronic villi biopsy

Chronic villus sampling is generally performed at 10-13 weeks. Placental villi may he obtained through transcervical. transabdominal or transvagmnal access to the placenta.

Advantage Results are available earlier in pregnancy which decreases parental anxiety when results are normal and allows earlier and safer methods of termination when abnormal.

Contraindications Vaginal bleeding or spotting, extreme ante or retrovcrted uterus and patients body habitus precluding easy access to uterus or clear visualisation of its contents with USG. Active infection is also a contradiction.

9. Percutaneous umbilical cord blood sampling (PUBS)

(CORDOCENTESIS)

Umbilical vein is punctured under direct ultrasound guidance, usually at or near its placental origin and blood is withdrawn and analysed.

Uses Primarily for assessment and treatment of confirmed red cell or platelet allo immunization

- analysis analysis of nonimmune hydrops
- karyotype analysis of fetal blood cells
- Metabolic and hematological studies
- Acid base analysis
- Viral cultures
- Immunological studies
- Congenital malformation or severe IUGR detection

Complications Umbilical cord vessel bleeding

- Hematorna
- Fetal - maternal haemorrhage
- Fetal bradycardia
- Abortion

10. Fetal tissue biopsy

In this prenatal diagnosis can he performed by direct analysis of fetal tissue obtained by USG guided skin or muscle biopsy.

Evans and colleagues (1994) used muscle biopsy to diagnose muscular dystrophy and mitochondrial myopathy. Elias and co-workers (1994) used skin biopsy in 17 fetuses at high risk for epidermolysis bullosa.

11. Preimplantation diagnosis - Polar body analysis. Blastomere biopsy This allows selection of only healthy embryos to avoid pregnancy termination.

a. Polar Body analysis

First polar body is expelled anyway and its removal for analysis Should not affect fetal development.

b. Biastomere biopsy

Utilizes 3 days old embryo (6-10 cell stage)- taken through a hole made in the zona pellucida. Loss of one totipotent cell at this stage supposedly has little or no effect on developing embryo.

Uses Diagnosis of single gene disorders such as cystic fibrosis or sickle cell disease. Sex determination in X linked disease and identification of aneuploidy with advanced maternal age or parental chromosome rearrangements.

12. Analysis of fetal cells in maternal circulation

All women have atleast small number of fetal cells in their bloodstream. Fetal cell isolation can be done by cell-sorting techniques. They can be separated by density gradient or protein separation techniques, fluorescence activated cell sorting or magnetic activated cell sorting. Nucleated red blood cells are most easily identified.

Uses

- Autosomal recessive diseases like thalassemia
- Fetal red cell D antigen typing
- Karyotyping using fluorescence in Situ hybridization (FISH)
- Screening test for aneuploidy

13. Ultrasound

Since the first obstetrical application of ultrasound imaging by Donald and Co-workers (1958), ultrasound evaluation of the pregnant uterus has become indispensable. Technical development in ultrasound equipment such as grey scales and real time imaging together with reasonable cost of many real time machinery has lead to widespread use of USG examination in pregnancy. Among the different modalities available. for detecting anomalies, USG has been the most effective and safe method.

BASICS OF ULTRASOUND

In ultrasound, ihc picture displayed on the screen in produced by sound waves reflecting from the imaged structure.

A sound wave is a mechanical disturbance propagating through a liquid, solid or gas medium. Sound waves are produced by vibrating sources that are in contact with the medium. Vibrations of source produce vibration of adjacent molecules in the medium, which in turn produce vibration of more distal molecules and so forth. The acoustic disturbance propagates away from the source at the speed of sound in medium. Speed of sound in air is about 330 m/sec. The effect of sound wave in air particles at any time will be associated with 'compressions' and 'rarefactions' of air molecules. In compressions, air molecules are squeezed together and in rarefactions they are drawn apart at periodic places in the medium. A plot of this pressure changes in the medium traces out a ‘sine wave’.

Speed of the sound wave is the distance traveled per unit time. Sound waves travel through tissue and other media at a speed that depends on properties of the medium, specifically the elasticity (or stiffness) of the medium and the density govern the propagation speed. The speed of sound is important because if its known, then the distance to any object can be computed using the time it takes for a sound pulse to travel to the object and an echo to return. USG is used to

image the soft tissue regions of the body. Average speech of sound in soft tissue is about = 1540 m/sec.

Longitudinal wave

When sound wave travels through air, water or soft tissue, the particles in the medium vibrate parallel to the direction of propagation of the sound wave. This is referred as 'longitudinal' vibration or 'longitudinal wave'.

Transverse wave

In some media, it's possible to generate sound waves in which the direction of vibration of particles is perpendicular to direction of travel of the wave - Transverse sound waves. These are created in solid materials (eg) bones in body. But these waves do not propagate through soft tissue.

Diagnostic ultrasound transducers are designed to produce ongitudinal sound waves as only these can be transmitted through soft tissues.

Physics of Ultrasound

The frequency of a sound wave is determined by the number of oscillation per second made by the vibrating source. Sound waves whose frequencies are greater than 20 kHz are referred do as ultrasonic. For medical diagnostic application, frequencies in the range of 1-10 mega hertz Million HZ. are commonly employed for the required resolution.

For the high frequencies and directional beams required in medical ultrasound, the reverse piezoelectric effect is used. Piezoelectric effect was first discovered by Pierre curie in 1880 when he found that mechanical stresses applied to a quartz crystal created a minute electrical potential across faces of the crystal. In reverse effect, by applying silver to the faces of a crystal and applying an electrical charge, he could create a mechanical change in the size of the crystal. Thus by applying electrical potential to a phase of the crystal sound waves could be created across when crystals are compressed. This phenomenon is used in the production of ultrasound and for convenience synthetic ceramic crystals are used instead of quartz. This transducer material when activated by electrical pulse starts emitting number of sound wave cycles.

When the alternating current is applied to a transducer made of piezoelectric material, intermittent high frequency sound waves exceeding 20.000 cps are generated. Applying a coupling agent, such as a water-soluble gel, diminishes the loss of ultrasound waves at the transducer skin interface. The transducer emits a pulse of sound waves that passes through the layers of soft tissue. When an interface between structures of different tissue densities is encountered some of the energy is reflected back to the transducer. The amount of energy reflected is proportional to the difference

in densities at the interlace. The reflected energy generates a small electrical voltage that is amplified and displayed on a screen and appears as a shade of color or brightness somewhere on the continuum between white and black. Bone is dense (echogenic) and generates a voltage that appears white on the screen, while fluid (anechoic) appears black. Soft tissues appear as varying shades of gray.

Ultrasonic Transducers

Transducer refers to any device that converts energy from one form to another. An ultrasonic transducer is the link between an ultrasound imaging instrument and the patient being examined. It is both the source and detector of USG signals and is a major factor in the overall performance of an instrument.

i. Linear - array transducer

These give a side coverage of area scanned, the rectangular area beneath the transducer and hence is good for scanning the abdomen for obstetric indication and medical disorders. Presence of hones, such as ribs and pubic symphysis will limit the acoustic imaging by linear probe.

ii. Sector - scanning

The advantage with sector scanning is that a point contact is made with the patient and this small acoustic window facilitates divergence of sound waves and image the objects in depth. Hence intercostals scanning

(echocardiography) and pelvic scanning are best done with sector scanners.

Transabdominal scanning is most commonly performed with a 3.5-7mhz curvilinear transducer which uses sequential firing of multiple crystals and generates an image so fast (more than 40 frames / sec) that the picture on the screen appears to be moving in "real- time". Higher frequency transducers yield better image resolution however lower frequencies afford better tissue penetration.

Display of Echoes (types of images)

1. A mode (Amplitude mode)

   Unidimensional vertical deflection from base line proportional to the strength of the echo generated.

2. B mode (brightness mode)

   2D image of the object made up of varying density of dots is obtained based on grey-scale quantification of echo signals.

3. BM mode (brightness Motion mode):

   B mode in motion. also known as real time scan.

4. TM mode (time motion mode)

   Gives the rate of motion per unit time. Used mainly for cardio Vascular study (ie) to compute FHR.

5. Doppler ultrasound:

Methods of assessment of movement by ultrasound.

Types of Ultrasound

LEVEL I

Expected only to establish fetal viability, gestational age and to detect multiple gestation. Also called basic / office/ low risk *I* screening ultrasound.

LEVEL II

Includes findings of level I + to record fetal anomalies known to be associated with elevated MSAFP. Also known as targeted / high detail / genetic / referral *l* fetal ultrasound.

LEVEL III Level II USG along with fetal echocardiography

LEVEL IV Interventional ultrasound.

3D

3D USG allows baby to be examined in all three dimensions

- can be performed at any gestational age.

- more accurate and baby can be visualised by parents.

4D

Not only allows 3 dimensional images but it can uniquely he done in real time.

Allows movemcnt and behaviour of baby to be studied.

Safety of USG

No confirmed biological effects in mammalian tissue have been demonstrated in the frequency range of medical ultrasound. (American Institute of Ultrasound in Medicine. 1991). In the low —intensity range of gray-scale imaging. no Fetal risks have been demonstrated in over 35 years of use(Maulik 1997: miller and Colleagues, 1998).

Indications for high resolution USG (Level II)

- Raised MSAFP
- Low MSAFP / positive triple test (MSAFP + HCG ± ESTRIOL
- Women with a personal *I* Family History of anomaly
- Oligohydramnios
- Polyhydramnios
- Maternal diabetes mellitus
- Women in preterm labour esp. if the presentation is breech.
- Women exposed to potential teratogens in early pregnancy.
- Women with suspicious findings on routine ultrasound.
- Patient with symmetrically small fetus.

FETAL ANOMALIES AND SCREENING BY USG

1. HEAD

a. Size and Configuration

Studied in BPD plane. CI to see doliocephaly / brachycephaly crianiostenosis (CI and BPD are persistently low)

b. Dimensions of lateral ventricles:

Studied in a plane above the BPD plane at 18 wks gestation which passes through the boides of lateral ventricles.

Lateral ventricular width (LVW) is the distance between midline echo and lateral wall of ventricle (generally anterior horn)

Hemisphere width (HW) is the distance between midline after 2^{nd} trimester (remains constant throughout life)

Normal LVW = 1/3 HW after 2^{nd} trim (remain constant throughout life)

After 18 wks, ratio – 0.5 (Normal upto 1)

i. Microcephaly

Small brain with in a small head.

- Head Circumference (HC) and Biparietal Diameter (BPD) are <3 SD from the mean for GA (single scan Duplex)
- Head Circumference (HC) / Abdominal circumference (AC) is very less

- Serial sonography - if HC and BPD are Very less as compared to AC and Femur Length (FL)

ii. Anencephaly

Absence of cranial vault and cerebra! hemispheres (detected from 12 wks gestation)

USG features

- Absence of cranial vault
- Short neck
- Typical-froglike appearance
- Polyhydramnios
- Sometimes a knot of echoes at the cephalic pole representing the base of the skull and orbits.

False positive rate is zero and as anencephaly is incompatible with life termination of pregnancy should be offered. There is always *5%* risk of recurrence in next pregnancy.

iii. Hydrocephaly

Ventriculomegaly due to abnormal accumulation of CSF. (ventricular dilatation precedes cranial enlargement by many weeks)

- Hydrocephaly with spinabifida occurs in 80% case
- Diagnosis can he made as early as 18 wks
- In practice a ventricle to hemisphere ratio is measured (VHR)

VHR=V/H x 100

Where V- Dist from midline echo to lateral border of lateral ventricle

H- Distance from midline echo to linear table skull.

Normal VHR *at* 16 wks GA = *55%* and after 24 wks 30%.

Hydrocephaly is diagnosed when BPD is 11 cm and AP diameter is 13cm. Hydrocephaly exhibits a multifactorial pattern of inheritance. After one affected pregnancy risk *of* second are of order of 1:20 and *after* 2 affected pregnancy, risk of further affected pregnancy are I in 10. Isolated hydrocephaly is more severe- may be due to aqueduct stenosis - sex linked recessive condition - recurrence rate I in *30.*

- Pit falls in diagnosing hydrocephaly

Choroids plexuses during glycogen phase is very echogenic in 11 trimester and fills the entire lateral ventricle posterior to Foramen of Munro and obscures lateral ventricle. At this time cerebral cortex is hvpoechoeic and may mimic appearance of dilated ventricles.

iv. Occipital meningocoele

Condition in which meninges protrude out as a sac.

v. Encephalocoele

Condition in which fetal brain has protruded into the sac.

vi. Shape of head

Banana sign curving of cerebellar hemispheres anteriorly with obliteration of cisterna magna, scalloping of frontal bones and traction on

Cerebellum forces it to wrap around the brain stern. (ARNOLD CHIARI malformation).

Lemon sign

Blunting of the sincipit or scalloping of the frontal hones, level same as BPD level.

2. FACE

Cleft lip / Palate

A cleft - lip defect results from lack of fusion between the median nasal and maxillary processes, an event which is normally completed hy35 days of intrauterine life. Cleft lip formation may also impair subsequent closure of the palatine processes (normally occurring by the 8^{th} to 9^{th} week of fetal life) resulting in a cleft palate. Approximately 70% of unilateral cleft lips are associated with cleft palate. UIL clefts are more likely to occur on left side and more frequently seen in males. Isolated cleft palate is a different entity seen in females commonly.

It's not fully determined whether the defect of cleft lip / palate is related to single gene or to multifactorial etiology. When both parents are unaffected recurrence risk is 2 - *5%.* If cleft is bilateral or if two siblings arc affected, risk

is 4- 10 %.

For ultrasound detection to visualizing cleft lip / palate -- 2 views are used. In first, angled coronal view of the face and in second lips and nares. Both views can be seen by line movements of the transducer in a position 45 ° - 90 ° from *BPD* plane.

Adjuncts in diagnosis of cleft lip / palate

- Undulating tongue movements
- Hypertrophied tissue at the end of the cleft and
- Hypertelorism

3. NECK

Nuchal skin thickening

Transverse section of fetal head which includes cerebellum and occipital bone. Its measured from the outer edge of occipital bone to Outer edge of fetal skin. Normal fat pad < 5mm in early second trimester. If> 5 mm between 15 - 20 wks gestation - trisomy 21.

Cystic Hygroma

Is a large fluid filled septated cysts extending from the posterior aspect of the neck. Congenital malformation resulting from jugular lymphatic obstruction- due to lack of communication between the cervical lymphatic vessels and the jugular venous system. Trapped lymphatic fluid accumulates, often causing

massive enlargement of the lymphatic spaces of the posterior aspect of the neck. When large cystic hygroma is identified prenatally by USG, fetal death inevitably occurs. Commonly occurs with turner's syndrome.

Criteria for prenatal sonographic identification of cystic hygroma are thin walled multiseptated. fluid-filed mass projecting posteriorly from the neck without evidence of a defect in the cranial vault and at a constant location with respect to occiput despite fetal motion. This is found in association with trisomy 13.18,21 and autosomal recessive disorders. Fetus with cystic hygroma and a normal chromosome analysis when identified, recurrence rate may be as high as *25%*.

4. CHEST

Diaphragmatic Hernia

Of the 3 types, most common is posterolateral form (hernia of foramen of Bochdalek). Its usually unilateral and commonly occurs in left side. It results from incomplete fusion of the pleuro-peritoneal membrane by 6-10 wks of fetal life.

II type Anteromedial (hernia of Foramen of Morgagni) results from maldevelopment of septum transversum in the retrosternal area. Pericardium may also be incomplete in which case hernia can he in pericardial cavity. In III type. Eventeration simulates diaphragmatic hernia Abdominal contents are found in the chest cavity secondary to thinning of portion of the diaphragm.

USG features

Mediastinal shift seen

abdominal organs seen in thorax

small abdominal cavity

Peristalsis of bowel is thorax

Polyhydramnios

5. ABDOMEN

In abdomen, Site of cord entry is important to check for the integrity of anterior abdominal wall. Stomach bubble is usually seen after 16 wks of gestation (transverse section).

Eosopagel atresia	:	Absence of stomach bubble
Duodenal atresia	:	Double bubble appearance.
Iejunal atresia	:	Triple bubble appearance
Omphalocoele	:	Middle abdominal wall defect with herniation of intestine / liver into the base of umbilical cord. Herniated viscera contained in a sac umbilical cord inserted into the apex of the sac. (generally seen in trisomies)

Gastroschisis : Right – side paraumbilical defect associated with evisceration of abdominal organs (generally an isolated defect).

Eosophageal Atresia

In normal fetus esophagus is not sonographicallv visible and is thought to he collapsed. But hypopharynx, larynx and trachea in fetal neck can be made out by USG after IS wks gestation.

In fetuses with mid-oesophageal atresia (the commonest type) and no communicating pathway with stomach, proximal oesophagus is transformed into a dilated pouch displacing the upper trachea anteriorly. In these fetuses the stomach 'bubble' in the abdomen would be lost and polyhydramnios develops. Although this triad is suggestive of oesophageal atresia, antenatal diagnosis is difficult irl 90% of such anomalies, as amniotic fluid reaches stomach by way of communication through trachea.

6. URINARY TRACT

Renal Abnormalities

It includes renal agenesis and renal dysgenesis which could be divided into infantile polycystic kidney, Meckel-Gruber syndrome, multi-cystic dysplastic kidney, congenital hydronephrosis and bladder outlet obstruction. Bladder outlet obstruction can he urethral agenesis. cloacal persistence, posterior urethral values

or urethral stricture.

1. Renal agenesis

Unilateral renal agenesis usually is asymptomatic- and have a normal life span. Associated genital abnormalities include uterine malformations (unicornuate or bicornuate or septate uterus), ipsilateral ovarian agenesis and atrophy of testes, prostate, epididymis and seminal vesicles.

Bilateral renal agenesis is rare and as fetal urination does not occur severe oligohydramnios is present. This restricts fetal movements within the uterus and leads to secondary compression deformities including clubfoot, joint dislocations and flexion contractures. Affected infants also have typical potter facies with prominent skin fold beneath the eyes. receding chin, flattened nose lungs that are incapable of meeting extra uterine ventilatory requirements. Death from asphyxia typically occurs within an hour of birth.

USG findings of bilateral renal agenesis

Severe oligohydramnios

Inability to visualize within the fetal bladder and

Failure to identify fetal kidneys

ii. Congenital Hydronephrosis

Caused by partial ureteral obstruction with secondary distension of' the renal pelvis and calyces. Narrowing may occur at either the uretcovesical or uretero-pelvic junctions, but latter is common. In USG, congenital hydronephnosis is characterized by a persistent. excessive collection of fluid within the renal pelvis.

Bladder Neck Obstruction

USG features

Dialted proximal urethra and distendend bladder (key – hole appearence)

Hydronephrosis / hydroureter

Oligohydramnious

Fetal bladder and abdominal enclargement may be significant enough to cause soft tissus dystocia and prevent vaginal delivery.

7.SPINE (Transverse / Longitidinal view)

Easily examined after 16 wks gestation. Normally a closed horeshoe shaped apperance of each verebra is maintanined (trensverse view)

Spina bifida / Menigomyelocoele

Refer to a defect in the spine resulting from failure of the two halves of the verebtral arch to fuse. Usually occurs is the lumbosacral and cervical regions. If the meninges protrude through this defect, the lesion is designated meningocoele; if the neural tissue is included it's a menigomyelocoele.

Normaly spine with an intact normal arch has the apperearance of a closed circle in transverse section while spina bifida appears as an open circle. In longitudinal sectors normally the skin covering of an intact spine

May be seen. In case of open spina bifida this is lost. Presence or adsence of associated hydrocephaly should also be noted.

USG features

a) Direct signs

- V shape of echoes from verebra
- Absence of skin and subcutaneous tissus over the effect

b) Indirect signs

- Associated with hydrocephaly / microcephaly / Iniencephaly
- Banana sign / lemon sign
- Rocker bottom foot
- Associated with meningocoele / meningomyelocele / myeloschisis
- Closed meningomyelocoele occurs in about 10% of patients with spina bifida

Caudal regression syndrome

Ranges from absence of sacrum with short femora to complete fusion of lower limbs (mermaid syndrome or sirenomelia)

Seen in infants of mother with IDDM

8.OTHERS

Fetal Hydrops

Is a state of excessive fluid accumulation into both the extra vascular compartment and body cavities, leading to devolopment of anasarca , ascites and pleural or pcricardial effusions. It is commonly dividend into 2 major categories (i) Isommune hydrops (ii) Nomimmune hydrops.

Isoimmune hydrops occur when there is material IgG antibody response to fetal RBC's. This is most frequently seen in Rh negative mothers sensitized previously and carrying a Rh positive fetus. The sensitized mother responds to her fetus by producing anti-D antibodies which cross the placenta and cause hemolysis of fetal Rh positive red cells.

Second group of non –immune hydrops (NIH) which develops due to a basis of pathologic process other than antigen –antibody RBC hemolysis.

Sonographic diagnosis of fetalhydrops

In hydrops fluid accumulation in some or all serous cavities and edema of all tissues is apparent. Placenta may also appear thick. Antenatal diagnosis is easy ascetic and pericardial fluid even when present in small quantities (25-100 ml) can be visualized ultrasonically. Additionally edema of fetal scalp and body is easily detectable.

SCAN SHOWING DUODENAL ATRESIA

9.CERTAIN MARKERS OF CHROMOSAL ANOMALIES

AND SYNDROMES

1.Duodenal atresia	-	Down's syndrome
2.Omphalocele	-	Trisomy 18
	-	Trisomy 13
	-	Triploidy
	-	Pallister – Killian syndrome
	-	Beckwith – wiedemann syndrome
3.Rocker – Bottom foot	-	Trisomy 18 (most frequent)
	-	Trisomy 13
	-	Caudal regression syndrome
	-	Diastrophic dysplasia
4.Polydactly	-	Trisomy 13
	-	Meckel – Gruber syndrome
	-	Orofacio digital syndrome Type II
	-	Carpenter syndrome
5.Choroid plexus cyst	-	Trisomy 18
6.Single umbilical artery	-	Renal malformations or renal agenesis
	-	Chrosomal aberrations
	-	IUGR

DETECTION RATE BY USG

Diagnosis	Pathology (defect)	Percentage of detection
Spina bifida	Open Spinal Cord	90%
Anencephaly	Absence of the top the head	99%
Hydrocephalus	*Excess fluid within the brain	60%
Major congenital heart problems		25%
Diaphragmatic hermia	A defect in the muscle which separates the chest and abdomen	60%
Exomphalos/gastroschisis	Defects of the abdominal wall	90%
Major kidney problems	Missing or abdominal kidneys	85%
Major limb abnormalities	Missing bones or very short limbs	90%
Cerebral palsy	Spasticity	Never seen
Autism		Never seen
Down Syndrome	May be associated with heart and bowel problems	About 40%

Supplement to ultrasound screening for fetal anomalies, July 2003

(Royal College of Radiologists)

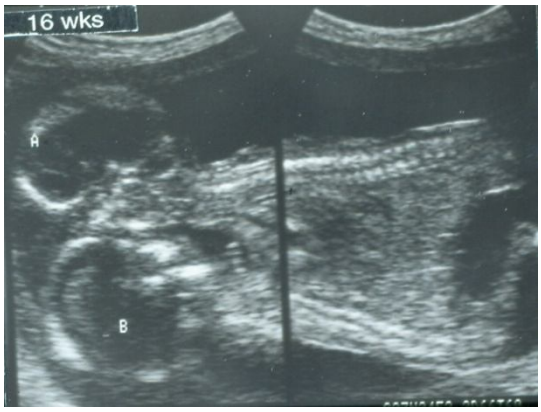
Conjoined Twins

- Incidence is 1:50,000 to 1:100,000
- Sporadic event caused by an incomplete division of the embryonic cell mass
- Different types of conjoined twins
    - Craniopagus – joined at the brain
    - Thoracopagus – joined at the heart
    - Omphalopagus – Xiphopagus – joined at the abdomen
    - Pygopagus – joined at the buttocks and lower spine

Ischiopagus – joined at the hips

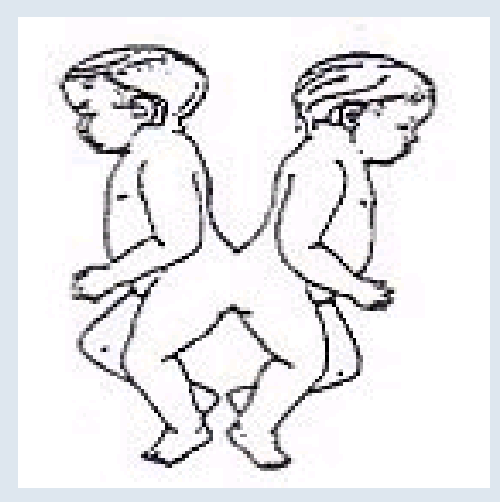
Thoracopagus

- Most common form of conjoined twins
- Congenital heart disease found in 75% of cases
- The union always includes the heart
- Most frequent abnormality is a conjoined heart with two ventricles and a varying number of atria



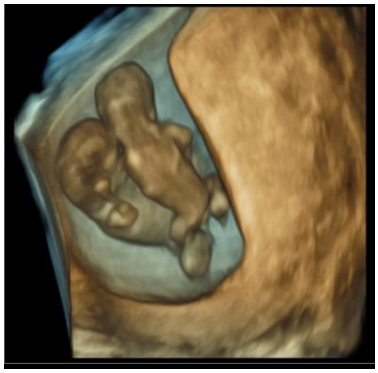
Pygopagus

- Joined at the buttocks and lower spine
- Face away from each other
- Have one anus, two rectums, four arms and legs



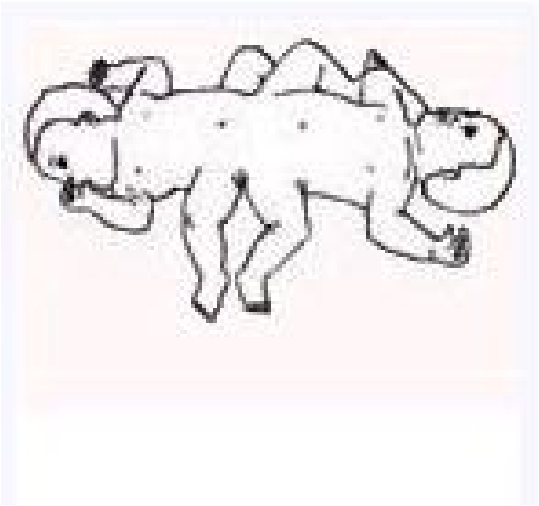
Omphalopagus in the first trimester

- Attached in the lower abdomen
- Remain facing each other throughout the exam



Ischiopagus

- Joined end to end with the spine in a straight line
- Four arms and a variable number of legs
- Have only one external genitalia



Fetal cystoscopy for thediagnosis and treatment of lower urinary outflow tract obstruction

Lower urinary outflow tract obstruction in a fetus may be associated with various developmental abnormalities. The obstruction may result from a number of pathologies, including urethral atresia or posterior urethral valves, and can be partial or complete. Severe obstruction may lead to oligohydramnios and pulmonary and/or renal dysplasia. If severe, pulmonary and/or renal dysplasia may cause death soon after birth from respiratory or renal failure, respectively, or the baby may require ventilatory support and/or renal dialysis or kidney transplantation. The long-term prognosis for children who require dialysis or transplantation in infancy is poor. Fetal cystoscopy is therefore indicated only if there is preserved kidney function. Alternative treatment options include expectant management, termination of the pregnancy, repeat vesicocentesis, open fetal vesicotomy or insertion of a vesico–amniotic shunt. Shunting aims to bypass the obstruction, with a view to definitive treatment of obstructive lesion(s) postnatally.

Single Umbilical Artery

Synonyms

Absence of an umbilical artery, umbilical artery agenesis, umbilical artery atrophy, and missing umbilical artery.

Definition

Single umbilical artery (SUA) is the absence of one Umbilical artery.

Epidemiology

Prospective studies indicate that SUA is present in 1 percent of all deliveries. The method of examination of the umbilical cord and the patient's race are important factors in determining the prevalence of this anomaly. For example, gross examination of the umbilical cord underestimates the prevalence. The location of the section is also important, since the two arteries may fuse close to the placental insertion of the cord and examination at this point would overestimate the prevalence of this anomaly. SUA is less common in Japanese and blacks and more common in eastern Europeans. SUA is more common in autopsy series and in stillbirths. The prevalence is higher in the third trimester than in very early embryos (less than 8 weeks old). This suggests that a developmental atrophy of a normally formed umbilical artery may occur in some fetuses. The male to female ratio is there is a greater tendency of males to be malformed in prospective series. The prevalence of SUA is three to four times higher in multiple gestations. There

is no evidence of an epidemiologic association between maternal age, parity, month of the last menstrual period, and the prevalence of SUA.

Etiology

There is no evidence of a familial tendency of this disorder. A genetic etiology is unlikely. The increased incidence in twin gestations is not observed in monozygotic twins. 14 The three theories about the pathogenesis of SUA are (1) primary agenesis of one of the umbilical arteries, (2) secondary atrophy of a previously normal artery, and (3) persistence of the original single allantoid artery of the body stalk. There is no statistical difference between atrophy and aplasia in SUA and associated malformations.1

Prenatal Diagnosis

The normal umbilical cord contains two arteries and one vein readily visible in transverse or longitudinal sections. In longitudinal sections, the helicoidal shape provides a typical braided appearance to the umbilical cord. A single umbilical artery can be seen readily in transverse sections by identifying a cord with only two vessels. The vein is typically larger than the artery. In longitudinal sections, a loss of the braided pattern of the umbilical cord can be visualized. Identification of an SUA is an indication for a careful search for associated anomalies including echocardiography. These infants are also at risk for intrauterine growth retardation, and serial examinations are recommended.

Associated Anomalies

Infants with SUA have a higher prevalence of congenital anomalies, intrauterine growth retardation, prematurity, and a higher perinatal mortality than infants born with two umbilical arteries. Twenty-one percent of infants with SUA have associated anomalies in prospective series, and the incidence is three times higher in autopsy series. Heifetz has estimated that the risk of anomalies is seven times greater in infants with SUA than in control infants with two umbilical arteries. It is clear that many of these anomalies are subtle (e.g., absence of the uvula) and, therefore, nondetectable with ultrasound. In other cases, the severity of the anomaly is such (e.g., bilateral renal agenesis) that identification of SUA is practically irrelevant. The mean number of malformations per infant varies between 2 and 5. Abnormalities that are detectable with ultrasound and are most commonly associated with SUA include cardiovascular abnormalities (particularly ventricular septal defects and conotruncal anomalies), cleft lip, ventral wall defects, esophageal atresia, spina bifida, central nervous system defects (hydrocephaly, holoprosencephaly), diaphragmatic hernia, cystic hygromas, genitourinary abnormalities (hydronephrosis, dysplastic kidneys), and digital abnormalities (polydactyly, syndactyly). All fetuses with SUA should have echocardiography performed, since cardiovascular abnormalities are among the most frequent defects. SUA is associated with a higher incidence of marginal and

velamentous insertion of the umbilical cord while these anomalies have been found in 5.9 percent and 1.2 percent of all placentas, respectively,30 in SUA, their incidence is 18 percent and 9.3 percent, respectively. In two different series, the association of SUA with velamentous insertion of the umbilical cord slightly increased the risk for other anomalies. The prevalence of chromosomal abnormalities in term infants with SUA is unknown. Isolated reports have documented that SUA can occur in association with autosomal trisomies.A recent pathologic study examining fetuses with SUA delivered before the 28^{th} week of gestation reported an incidence of chromosomal anomalies of 67 percent (6 out of 9). This is higher than the 31 percent (24 out of 74) observed in infants born with malformations other than SUA. In this small series (n = 9), all infants with SUA and chromosomal anomalies had severe malformations. From these data, performance of amniocentesis seems justified when SUA is associated with severe anomalies.

Prognosis

The mean perinatal mortality for infants with SUA has been reported to be 20 percent. Two thirds of the perinatal deaths are stillbirths, and among these, three quarters have occurred antepartum and one quarter intrapartum. The main cause of death is the presence of associated anomalies. However, the perinatal mortality remains elevated in infants with SUA but without associated malformations. This

is mainly due to prematurity and intrauterine growth retardation. Infants with SUA are at risk for internal malformation even if external anomalies cannot be detected. However, if these infants remain asymptomatic during the neonatal period, their risk for lethal or serious anomalies is not higher than that of non-SUA infants. The long-term prognosis for growth-retarded infants with SUA is good, since these infants attain growth rates comparable to non affected infants.

Obstetrical Management

The detection of a single umbilical artery should prompt a search for associated anomalies. Echocardiography is indicated. Karyotype determination should be performed if associated anomalies are detected. The risk of chromosomal abnormalities in SUA without gross anomalies detected by ultrasound has not been established. Serial sonography for identification of IUGR is recommended. Intrapartum fetal heart monitoring is indicated, since some series suggest that these infants are at risk for intrapartum fetal distress and death. Pediatricians should be alerted to the diagnosis of SUA, and noninvasive techniques like neonatal ultrasound should be used freely to detect subclinical anomalies. Invasive procedures for diagnostic purposes in an otherwise asymptomatic infant do not seem justified. Data from the Collaborative Perinatal project show that infants born with SUA had a higher incidence of inguinal hernias (5.5 percent versus 1.1 percent) than a control group in a follow-up period of 4

years. The IQ of nonmalformed infants with SUA is not different from that of infants with two umbilical arteries.

COUNSELING

Birth defects are the leading cause of death before infancy accounting for 20% of such deaths. 50% of spontaneously aborted fetuses have a chromosomal abnormality. Since many of these defects run in families and can recur in future pregnancies and since parents undergo major psychological trauma, counseling should be done for these patients.

Genetic counseling is a branch of genetics which deals with counseling of parents, potential parents and individual regarding genetic disorders for the birth of healthy baby free birth defects and inherited disorders.

Counseling is done to estimate any special extra risk of diseases malformation and handicaps in any children the couple may have, to put this into perspective for their child if he should prove to be affected the possibility and reliability of prenatal diagnosis, the availability of any curative and palliative treatment, information regarding alternative course of action and to reduce the birth frequency of genetically determined disease.

Genetic counseling should be done as early as possible to give opportunity to govern family size and less physical and psychological trauma to the family.

FETAL THERAPY

Include any intervention that may benefit a fetal condition diagnosed prenatally.

Before doing fetal therapy accurate diagnosis of malformation and its severity should be assessed and unnecessary intervention should be avoided.

Fetal therapy can be

- Medical  Mother
  Fetus
- Surgical

Medical

Prophylatctic Anti D can be given for Rh negative mothers Corticosteriods supports fetal adrenal gland s and prevent masculinizaion in congenital adrenal hyperplasia. Amnion infusion can be done in serve oligohydramnios to prevent constractures and anomalies.

Fetal Surgery

This is done for correction of structural fetal malformations. The prerequisite for surgery is to know in detail about the natural history of the condition probable outcome with or without therapy and the procedure should be tested successfully in experimental animals – primates.

1. Urinary shunts for obstructive uropathy

Uninary obstruction coult be due to posterior urethral vale or urethral atresia or ureteropclvic junction obstruction. Outflow obstruction will increase intrarenal pressure and cause dysplastic changes.

Renal cystoscopy can be done. Vesico- aminotic shuting – due to double J stent modified by double digital catherter.

Cystoscope guided laser excision can also be done.

2. Fetal Pleural effusion

Fetal chylothorax causes increase in perinatal morality. Aim is to prevent pulmonary hypoplasia, reversal of hydrops and thereby prolong pregnancy.

Criteria for treatment

Mediastinal shift with significant lung ompression

Abnormal Doppler patterns of great arteries before 35 wks.

Hydrops.

Treatment

Thoracocentesis

Thoraco – amniotic shunting

Pleuro – amniotic shuting – decompression of ches lung expansion.

Prevention

Low fat, high medium chain triglycerides in maternal diet to reduce the lymphatic flow.

3. Congenital Diaphragmatic Hernia

In this fetal surgery is done to allow pulmonary development and prevent pulmonary hypoplasia.

a) Plug

If liver herniaion is diagnosed before 25 wks – goretex patch can be applied to repair diaphragm. Plug the defect until the lung grows.

b) Tracheal Occlusion

By clips or inflatable balloon. It blocks the normal outflow of fetal lung fluid and stimulate of hypoplastic lungs in diaphragmatic hernia.

4. Sacrococcygeal Teratoma

Congenital highly vascular tumour. Treatment – Laser occlusion of selective feeding vessels.

5. Congenital Cystic Adenomeatoid Malformation and Pulmonary Sequestration (CCAMS)

CCAMS are pulmonary hamartomas consisting of overgrowth of terminal bronchioles. There are 3 types – Type 1,2,3. Features are mediastinal shift, pleural effusion, hydramnios and perinatal death.

Treatment – Lobectomy and Thoraco amniotic shunting

6. Neural Tube Defects

Lower spine defects covered endoscopically by maternal skin grafts (22 – 24 wks) prevent herniation of hind brain into foremen magnum.

Sealing of membrances : Intra amniotic injection of platelets and cryoprecipiate –PPROM

7. EXIT (Ex Utero Intraparturm Treatment)

To maintain fetal gas exchange until adequate ventilation is maintained. Its used is cases of potentially life threatening airway obstruction at birth in gaint fetal neck masses – lymphangiomic cervical teratomas.

MATERIALS AND METHOD

This study was conducted in the department of Obstetrics and Gynaecology at Government Rajaji Hospital,Madurai in the year 2013 June to 2014 July.About 150 patients examined perday in the outpatient antenatal clinic .Among them 200 antenatal patients who fulfilled the selection criteria were randomly selected and included serially for this study.

DATA COLLECTION

Age, Parity, Occupation, Socioeconomic status, duration of Marriage, Consanguinity, Previous history of abortions and still birth, History regarding drug intake and maternal infection, history of previous anomalous babies, level one basic ultrasound findings and level two special anomalous screening findings, therapeutic terminations, need for additional investigations, need for invasive procedures, mode of delivery and anomalies defected in abnormal non lethal anomalous scan findings.

Inclusion Criteria

I. Women with 18 -20 weeks of gestation willing to participate in this study with

1) Consanguineous marriage
2) H/O fetal anamolies in previous pregnancies
3) Patients with BOH
4) Polyhydramnios ,Oligohydramnios
5) Elderly gravid women
6) H/O drug intake radiation exposure during 1st trimester
7) H/O viral fever in 1st trimester
8) Patient is on treatment for medical disorder like epilepsy, diabete smellitus, congenital heart disease, bronchial asthma.
9) Threatened abortion with hormonal intake .
10) Rh negative mothers for whom anti D not given.
11) Those have done anamoly scan outside, having anamoly included in the study .Anamoly confirmed &managed accordingly.

Exclusion criteria

I. Those not willing to participate in this study

All normal antenatal patients having no risk factor mentioned above and having no signs of IUGR are excluded from the study.

After selecting the cases detailed history taking ,age, parity,occupation ,socioeconomic status, duration of marriage ,consanguinity, history regarding drugintake ,maternal infection, giving importance to previous history of anamolous babies ,or family history of congenital anamoly among blood relatives were taken. Through clinical examination was done, uterine size and the amount of liquor noted clinically. Routine investigations like urine analysis for albumin sugar deposits were done . blood investigations like Hb% ,grouping & typing ,blood urea sugar &serum creatinine were done .Then these patients were subjected to scan.Fetal biometry and detailed level II scan was done .If anamolies incompatible with life were detected ,patients were counseled regarding termination. If fetus with nonlethal anamolies were detected patients were counseled about the defect ,and the treatment options available after birth .Pregnancy allowed to continue and after delivery referral to pediatric surgery done. If fetus was normal, pregnancy continued and followed up.

AGE DISTRIBUTION

	No.of cases				
Age Distribution	Abnormal	Normal	Total	Abnormal %	Normal %
< 20	4	0	4	12.12	0.00
21 - 25	5	99	103	15.15	84.62
26 - 30	11	18	29	33.33	15.38
> 30	13	0	14	39.39	0.00
Total	33	117	150	100.00	

From this Table among 150 women taken for study 5 women were lessthan 20 years old. 102 between 21-25 years old. 29 between 26-30 years old. 13 between greater than 30 year old. 33.33% of women were between 26-30 years old, 39% of women were >30 years old. This shows the risk of anomalies more in older age group.

OBSTETRIC CODE

	No.of cases				
Obstetric code	Abnormal	Normal	Total	Abnormal %	Normal %
Primi	16	77	93	48.48	65.81
Multi	17	40	57	51.52	34.19
Total	33	117	150		

Among 150 women studies : Primi - 93

Multi - 57

Among the primi 48.48% had anomaly 65% normal

Among the multi 51.52 had anomaly 34.19% normal study.

Primi vs Multi (48.48% vs 51.52%). This equality may be because of elderly primis having more anamolies.

BOOKING HISTORY

	No.of cases				
Booking	Abnormal	Normal	Total	Abnormal %	Normal %
Booked	26	114	140	78.79	97.44
Unbooked	7	3	10	21.21	2.56
Total	33	117	150		

In this study : 140 booked cases

10 booked cases

21.21% unbooked cash had anamoly compared to 97.4% normal study were booked cases

CONSANGUINITY

	No.of cases				
Consanguinity	Abnormal	Normal	Total	Abnormal %	Normal %
II CM	18	11	29	54.55	9.40
III CM	8	103	111	24.24	88.03
NCM	7	3	10	21.21	2.56
Total	33	117	150		

Among the 150 women studied 10 had NCM, 140 had consanguineous marriage. 29 had II^0cm, 111 had III^0cm. In II^0 consanguinity 54% had anamolies. In III^0 24% had anomalies. 21% NCM marriage mothers had anomaly.

This shows increased risk of anomalous babies: with Consanguineous marriage.

CM vs NCM P Value - 0.011

statistically Significant

FAMILY HISTORY

	No.of cases				
Family History	Abnormal	Normal	Total	Abnormal %	Normal %
S	0	0	0	0.00	0.00
H	0	0	0	0.00	0.00
Sibling	3	0	0	9.09	0.00

Of These 33 patients with anamolous babies 3 of them already given birth to anamolous babies. Recurrence of anomaly in 9% of cases.

PAST HISTORY

	No. of cases				
Past history	Abnormal	Normal	Total	Abnormal %	Normal %
DM	1	0	1	3.03	0.00
Epilepsy	1	4	4	3.03	0.00
Heart Diseases	1	5	6	3.03	4.27
Viral	2	0	2	6.06	0.00
HT	0	3	3	0.00	2.56

1. Among the 150 women studied 1 had GDM – that mother had anomaly 3%. 4 had epilepsy, 1 mother had anomaly, 6 mothers had heart disease. Of them 1 had anomaly.
2. 2 Patients had previous H/O Viral illness both had anomalies.
3. 3 Patients had H/O, PIH → none of them had anomalies in this study.

PRESENT OBS. HISTORY

	No.of cases				
Prev. Obs. History	Abnormal	Normal	Total	Abnormal %	Normal %
Oligo	3	7	10	9.09	5.98
Poly	8	2	10	24.24	1.71

Among the 150 women studied 130 had normal amniotic fluid volume among this 22 had anomalous babies (16%). 10 Patients had oliohydramnios of these 3 had anomalies (9%). 10 Patients had poly hydramnios of these 8 had anomalies (24.24%).

SOCIO ECONOMIC STATUS

	No.of cases				
Socio economic status	Abnormal	Normal	Total	Abnormal %	Normal %
Middle class	11	110	121	33.33	94.02
Lower class	22	7	29	66.67	5.98

Anomalies more common in (66.67%) in lower socio economic status group.

NUTRITIONAL DEFICIENCY

	No. of cases				
Nutritional Deficiency	Abnormal	Normal	Total	Abnormal %	Normal %
Present	19	2	21	57.58	1.71
Absent	14	115	129	42.42	98.29

Anomalies more common in Patients presented with signs of nutritional deficiency.

CONTINUATION / TERMINATION

C/T	No. of cases	Percentage
C	3	9
T	29	87
Nil	1	3

1 case lost follow up. Pulmonary and Tricuspid atresia with large VSD.

1. TOF with small size pulmonary artery - Alive 6 months old
2. Choroid plexus cyst with Bifrontal Scalloping - Edward Syndrome – died at 28 days of life.
3. Cystichygroma of neck died at 10 days of life.
4. 29 cases of lethal anomalies were terminated.

ANOMALIES AND THEIR OUTCOME

Anomalies Detected	No.	Termination	Continuation
Arnoldchiari malformation	1	✓	
TOF C Small size PA	1		✓
Congenital Diapragmatic hernia	2	✓	
Anencephaly	4	✓	
Nonimmune hydrops	1	✓	
Pulmonary &Tricuspid atresia with large VSD	1		
paritel encephalocele c hemivertibrae	1	✓	
Choroid plexus cyst with Bifrontal scalloping occipital flattening	1		✓
Bilateral Renal Dysplasia	1	✓	
Mesomalic Type Of Skeletal Dysplasia	1	✓	
Cystic Hygroma	1		✓
Bladder Outlet Obstruction	2	✓	
Meckel Gruber Syndrome	1	✓	
Limp Body Wall Complex	2	✓	
Iniencephaly	1	✓	

Gastrochisis	1	✓	
Heterotaxy Syndrome	1	✓	
Rocker Bottom Foot	1	✓	
Uretheral Atresia	1	✓	
Craniochisis	2	✓	
Twins With Coarctation Of Umbilical Cord Of Single Fetus	1	✓	
Conjoint Twins	4	✓	
Amniotic Band Syndrome	1	✓	
TOTAL	33		

Mother with this anomaly (Pulmonary &Tricuspid atresia with large VSD) advised termination. she had lost from follow up.

DISCUSSION

Regarding age distribution it is evident that there is an increased chance of scongenital anamolies as maternal age increases. In this study higher percentage of anamolies were found in those > 30 years .39 % (13/33) anamolies in those > 30 years. Studies from simpson Sl,Golbus MS Martin AO et al,Genetics in OBG,New YorkGrune and Straffon also show that the risk of anamolies increase with increased age group.

Regarding parity distribution ,in this study primigravida woman had merely equal number of anamolies (48.48% vs51.52%) in multi.This equality may be because of elderly primis having more anamolies.Nicolaides KH,Campbell S,Gabbe SH et al.

It universally accepted that consanguineous marriages are at increased risk of anamolous babies .Nadiri S :Congenital abnormalities in newborns consanguineous and nonconsanguineous parents.conguineous unions are at increased risk to produce children with Autosomal Recessive disease also at increased risk to offspring with multifactorial condition,becse of potential for shared deleterious genes .In this study II nd &IIIrd degree Consanguinity had increased malformation (IInd -54%,IIIrd- 24%)

According to Friere _Maia N ,First degree relatives share hslf their genes ,second relatives share a fourth and third degree relatives share one eighth.

In this study among the 8 who has previous anamolous babies 5 had repeated anamolous babies this time also .Among these 5 mothers 1 patient had previous anencephaly ,had anencephaly this time also and was terminated .This patient was prescribed folic acid after 1 st loss but she has not taken it .The recurrence risk for neural tube defect after one affected child is 3-4 % and after two affected children it is 10 %.With folic acid supplementation risk after one affected child decreases by 70% to <1% .Czeizel AE ,Dudas I:Prevention of the first occurrence of neural tube defects by periconceptional vitamin supplementation.

Considering the amount of liquour and its association with congenital anamolies,In this study among 10 woman who has poly hydramnios 8 woman had congenital anamolies ,and among the 10 woman who has oligo hydramnios 3 had congenital anamolies .Incidence is 24.5 % & 9% respectively .Furman and coworkers ,perinatal mortality rate increased ,and fetal death complicates up to 40 % of pregnancies with hydramnios and anamolous fetus .

Toubal and collegues 2007 also states increased risk of anamolies with hydramnios .In a study by Biggio Jr ,Hydramnios prediction of adverse perinatal

outcome 20% of cases of polyhydramnios had associated fetal anamolies .This study agrees with that of Biggio et al.

Among the 150 women screened 10 % women with medical disorder was taken for the study .Of these 1 % women with anti epileptic drugs one had occipital encephalocele wiyh hemivertibrae .North Pacific epilepsy Research shows that exposure to antiepileptic drug increases risk of anamolies.Kultionek et al study at Uppasalo University Sweden shows that antiepileptic drugs increases the risk of neural tube defects .

Incidentally one woman with HIV positive presents with twin gestation with single fetal demise due to coarctation of umbilical cord.

Regarding booked & unbooked cases 21.2% of unbooked cases had anamoly .

Regarding, socioeconomic status and nutritional deficiency,those from low socioeconomic status and with signs of nutritional deficiency 57 % had anamolous babies compare to others.

SUMMARY

This study was conducted in the Department of Obstetrics and Gynaecology ,Government Rajaji Hospital Madurai during the period from June 2013 to July 2014

150 antenatal mothers were randomly selected from the patients attending the antenatal op strictly following the inclusion and exclusion criteria mentioned before ,and level II sonogram was done to detect congenital and acquired anamolies .

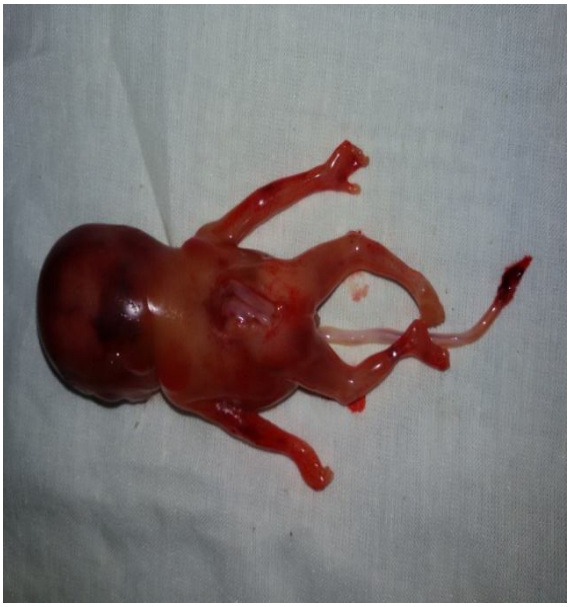
Proper counselling was done to the parents showing the anamolous babies and explaining the type of anamoly and its outcome .The effect of counseling and the management of mothers with anamolous babies were recorded.The results are tabulated and analysed .The following are the summary of the study.

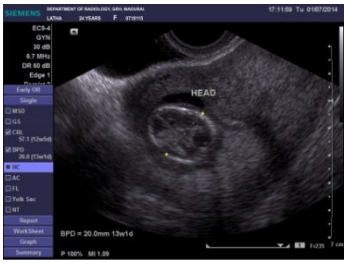
- Majority of the study group belongs to the reproductive age group 20-25:48.48%) (26-30:33.33%)
- 30 years old =39 % had anamolous babies
- In Reproductive age group 90 % had normal babies showing the effect of maternal age in congenital anamolies and the safety of pregnancy in reproductive age group.
- 21.2% unbooked cases had anamoly.

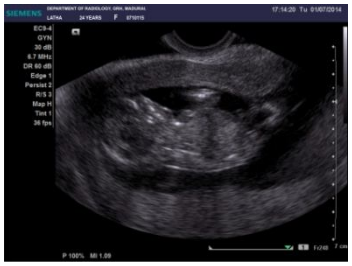
- 62% of patient belonging to this study were primigravida. 38% of mutipara constitute the study
- On critical evaluation of congenital anomalies with parity incidence of anomalies found to be same in primi 48.48% multi 51.52. There by showing parity has no effect over the incidence of congenital anomalies.
- Correlating consanguinity with congenital anomaly it's found that 54.55% II^0 cm patients and 24.24% III^0cm patients had anomalous babies.
- Incidence of congenital anomalies were directly proportional to the degree of consanguinity 54.55% Vs 24.24% in II^0 cm and III^0cm.
- Evaluation of the previous risk factors in the study revealed the importance of the previous fetal anomaly with congenital anomaly.
- When mother had a previous fetal anomaly recurrence rate was 40%.
- This study reveals that 1/4th of the pregnancy with abnormal amniotic fluid volume showed congenital anomalous babies. Congenital anomaly rate was 25% in poly hydramnios.
- Correlating medical disorder with congenital anomaly study showed epilepsy with antiepileptic drugs being associated with congenital anomaly. One case presented with occipital encephalocele with hemivertebrae.
- Congenital anomaly can be 100% avoided in diabetic pregnancies when properly treated and have a good glycemic control in the antenatal period.

- 87% lethal anomalies were induced and terminated with the consent of the mother. Reducing the maternal morbidity, psychological burden to the mother and work load to the health care personnel.
- 9% Non lethal anomalies followed up.
- TOF with small size pulmonary artery - Alive 6 months old
- Choroid plexus cyst with Bifrontal Scalloping - Edward Syndrome – died at 28 days of life.
- Cystichygroma of neck died at 10 days of life.
- 3% (1) case lost follow up. (Pulmonary and Tricuspid atresia with large VSD).
- This study reveals the congenital anomalies incidence at random selection in high risk group is 22%.

SCAN AND PHOTO SHOWING ARNOLD CHIARI MALFORMATION – II

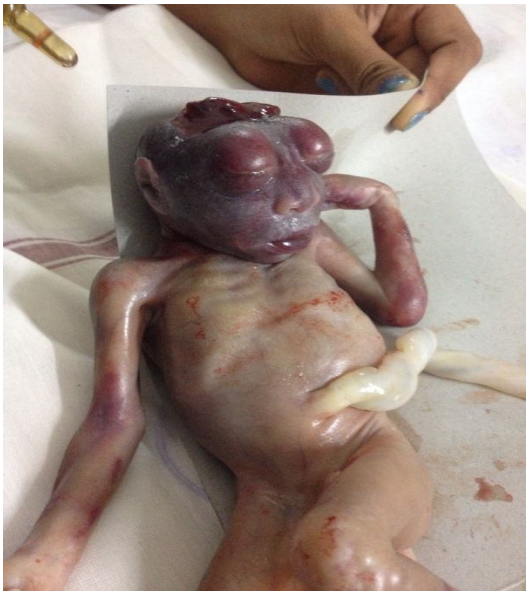


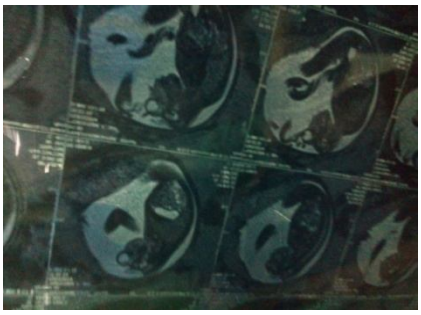




Banana and lemon sign are produced by caudal displacement of Cerebellar vermis fourth ventricle and medulla constituting Arnold Chiari Malformation – II.

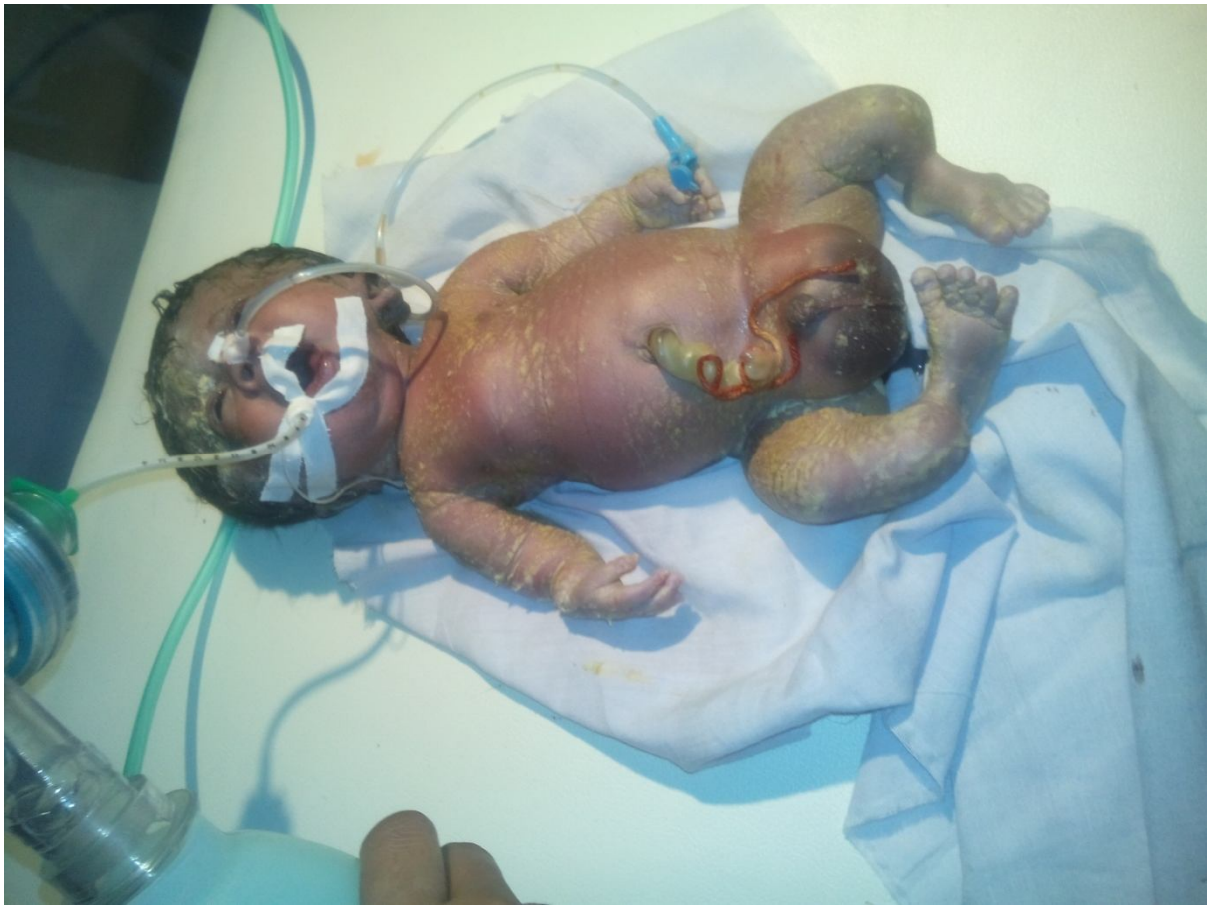
SCAN AND PHOTO SHOWING ANENCEPHALY





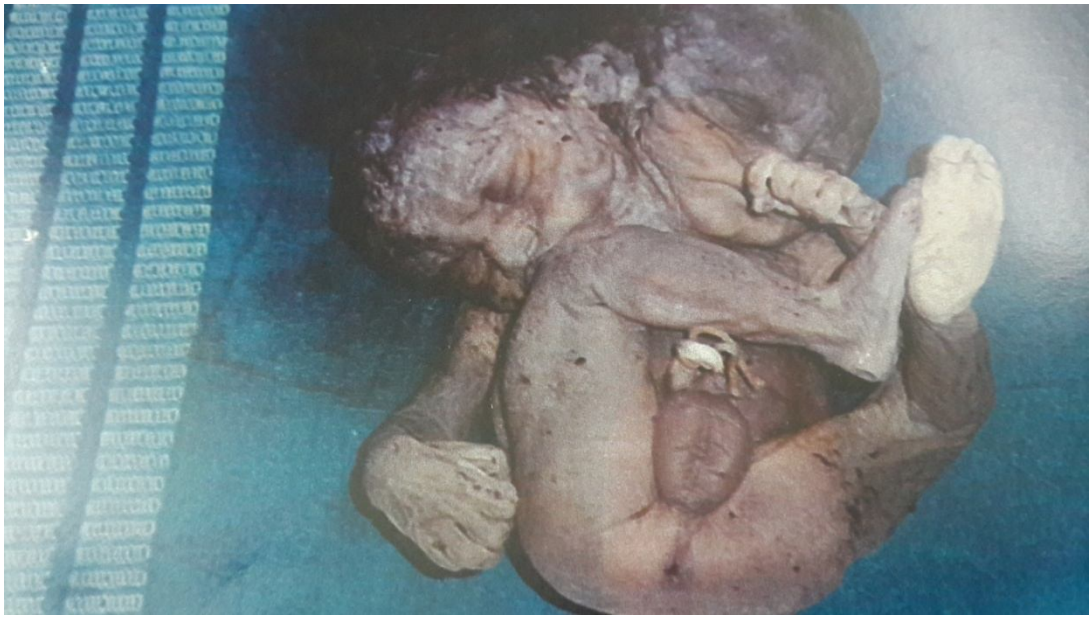
Anencephaly is a lethal defect characterized by absence of brain and cranium above the base of skull and orbit .It is the most common type of neural tube defect.

PHOTO SHOWING MESOMALIC TYPE OF SKELETAL DYSPLASIA

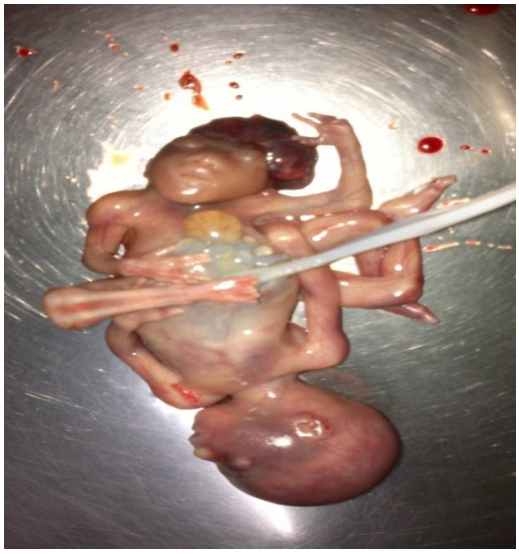


Clinically and radiologically the disease is characterized by severe shortening of long bones (limb's both proximal and median segments are affected), aplasia or severe hypoplasia of ulna and fibula, thickened and curved radius and tibia. These anomalies can cause deformities of the hands and feet. Hypoplasia of the mandible can also be present.

PHOTO SHOWING CONJOINED TWINS

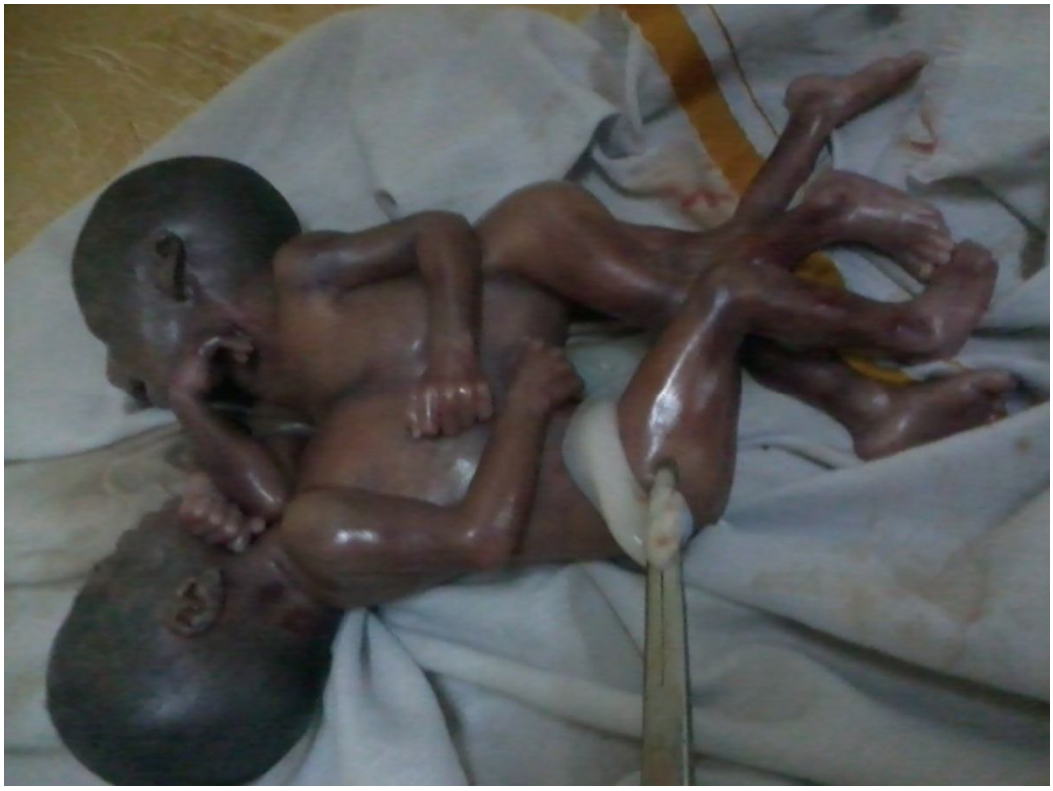


Dicephalic Diprosopic Parapagus - Mono amniotic monochorinic twins.



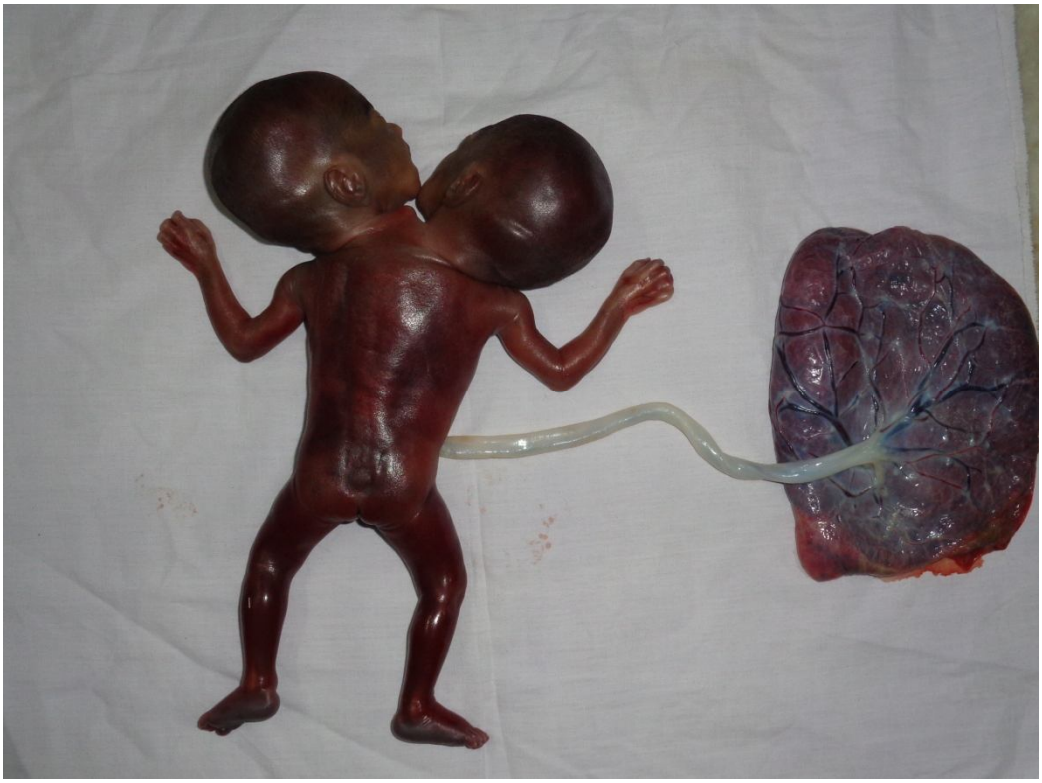
Conjoined twins with Ischio Pyophagus.

PHOTO SHOWED CONJOINED TWINS



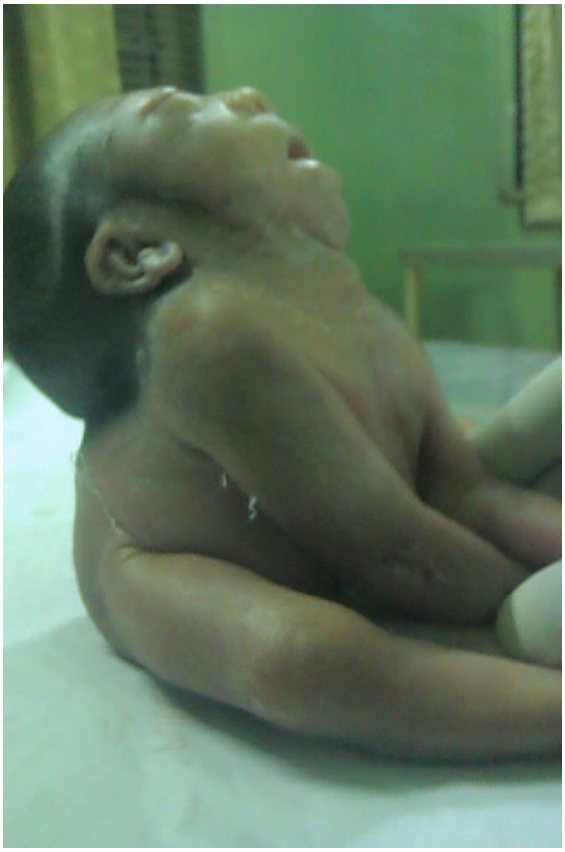
Thoracophagus omphalophagus mono amniotic mono chorionic. Both female baby. Twins joined at thorax & abdominal region.

PHOTO SHOWED CONJOINED TWINS



Paraphagus two head, two neck, two vertebral columns demonstrated.

PHOTO SHOWING INIENCEPHALY

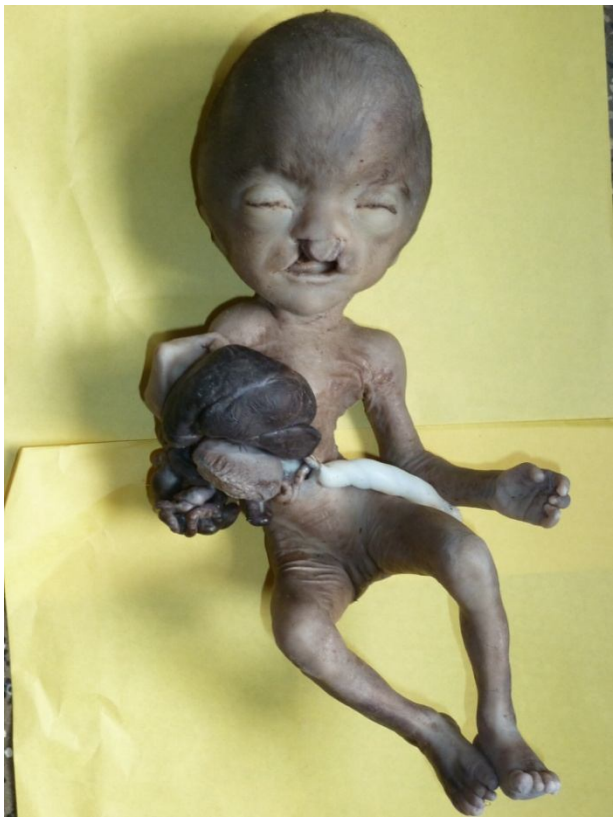


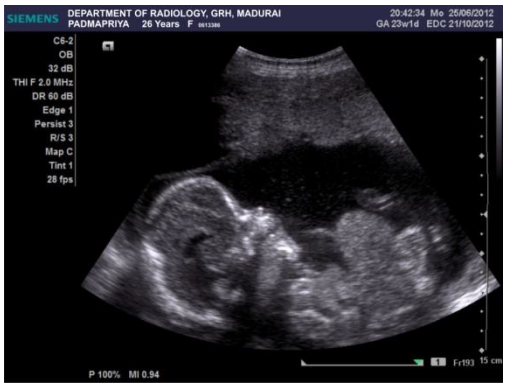
Iniencephaly, a term derived from the Greek word "inion" for nape of the neck, is a rare type of cephalic disorder that was first described by Étienne Geoffroy Saint-Hilaire in 1836. Those afflicted with the disorder all share 3 common characteristics: a defect to the occipital bone, spina bifida of the cervical vertebrae and retroflexion (backward bending) of the head on the cervical spine. Stillbirth is the most common outcome, with a few rare examples of live birth, after which death almost invariably occurs within a short time.

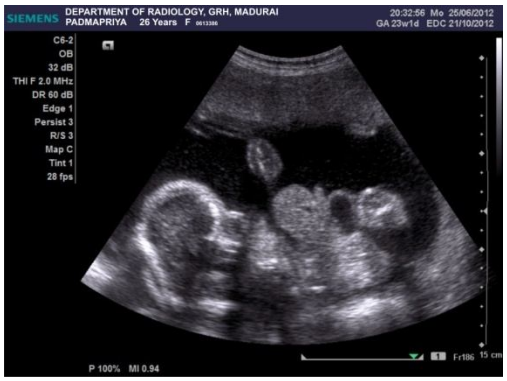
PHOTO SHOWING EDWARDS SYNDROME – ROCKER BOTTOM FOOT

Edward syndrome - Trisomy as an overall frequency of 1 in 8000 newborns and is three to four times more common in females. Striking facial features include prominent occiput, rotated and malformed ears, short palpebral fissures, and a small mouth.

PHOTOS SHOWING LIMB BODY WALL COMPLEX

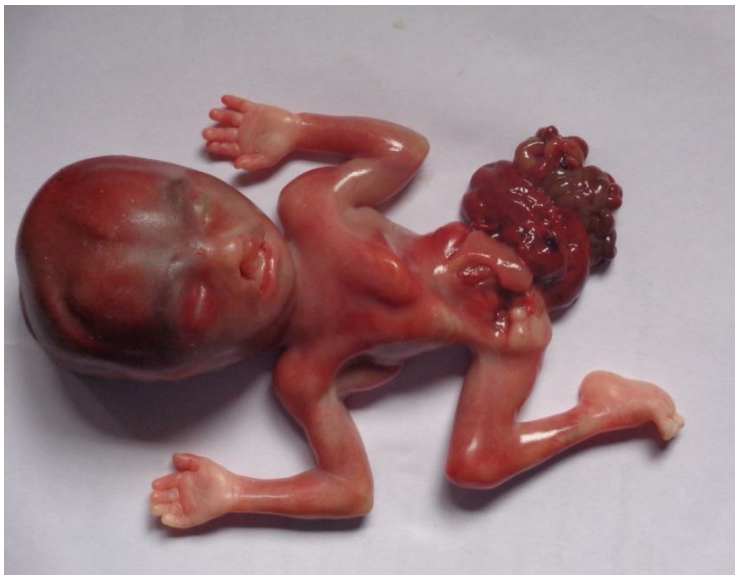


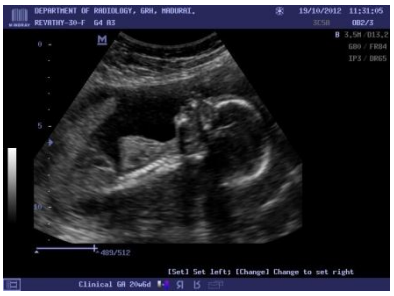


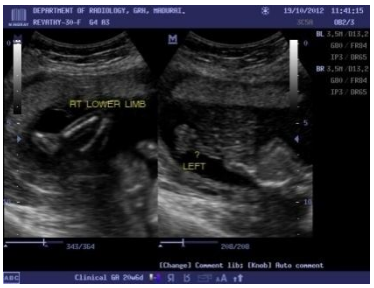


Anomaly consist of a poly malformation syndrome with a thoraco & or abdomino schisis associated with an eventration of the internal organs and anomalies of the extremities.

PHOTO SHOWING LIMB BODY WALL COMPLEX

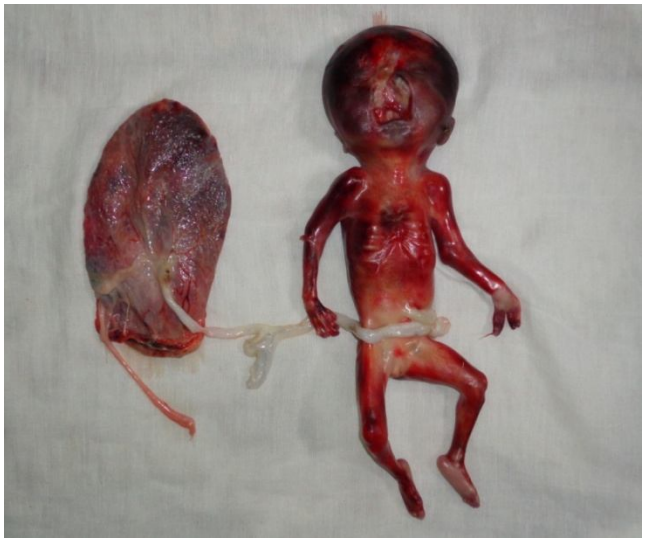


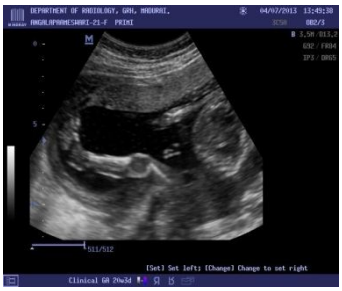


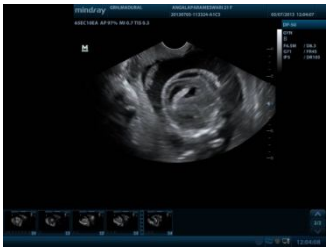


Anomaly consist of a poly malformation syndrome with a thoraco & or abdomino schisis associated with an eventration of the internal organs and anomalies of the extremities.

PHOTO & SCAN SHOWING AMNIOTIC BAND SYNDROME



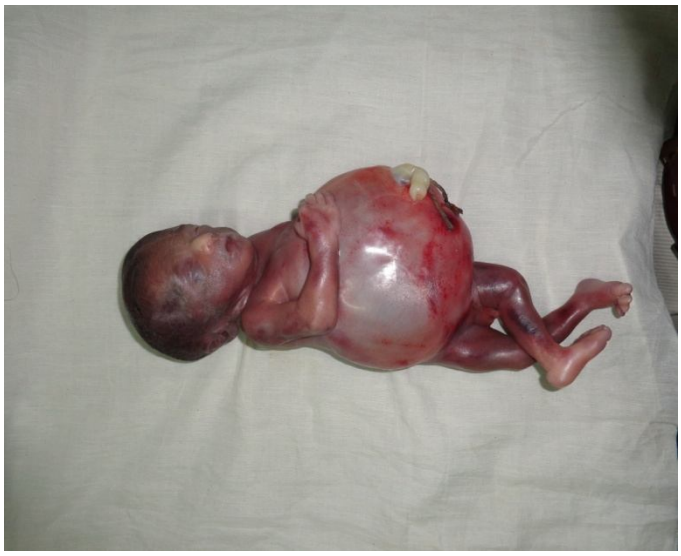


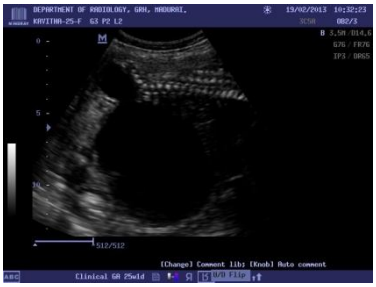


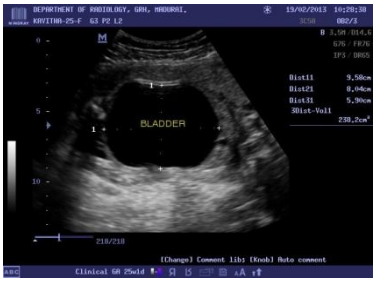
Amniotic Band Consist of fibrous strands extending from the outer surface of chorion in to the amniotic havity.

ABS is a well defind clinical entity presenting with deformities limb, thorax, craniofascial, skeleton, soft tissue and umbilical cord in a nonembryonic distribution.

PHOTO & SCAN SHOWING BLADDER OUTLET OBSTRUCTION

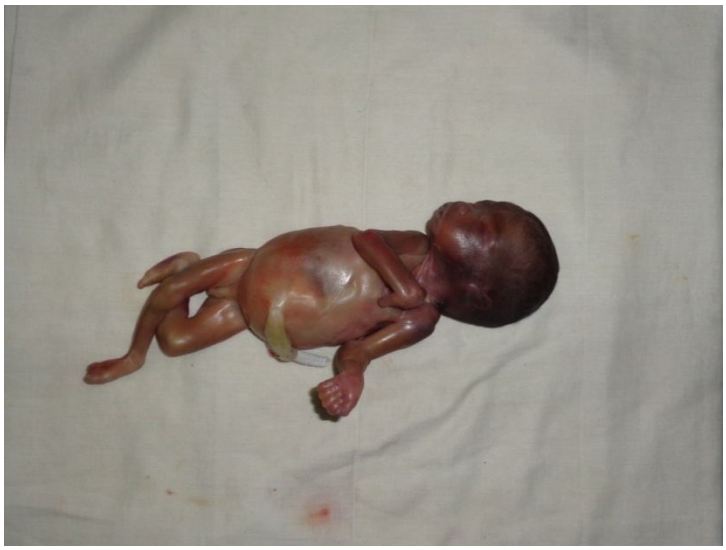


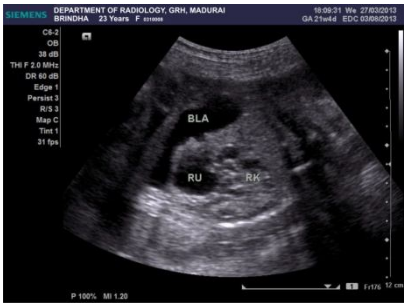


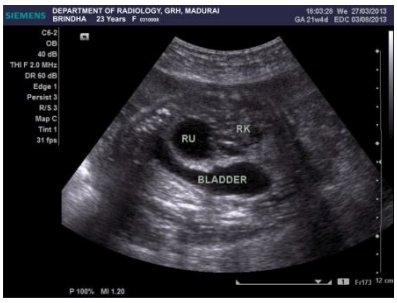


This distal obstruction of the urinary tract is more common in male fetuses, and the most common etiology is posterior urethral valves. Characteristically, there is dilatation of the bladder and proximal urethra.

PHOTO & SCAN SHOWING BLADDER OUTLET OBSTRUCTION

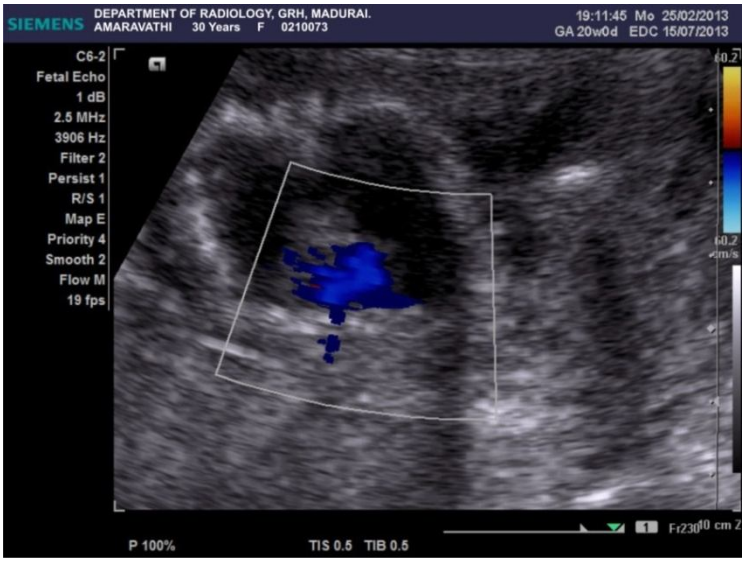


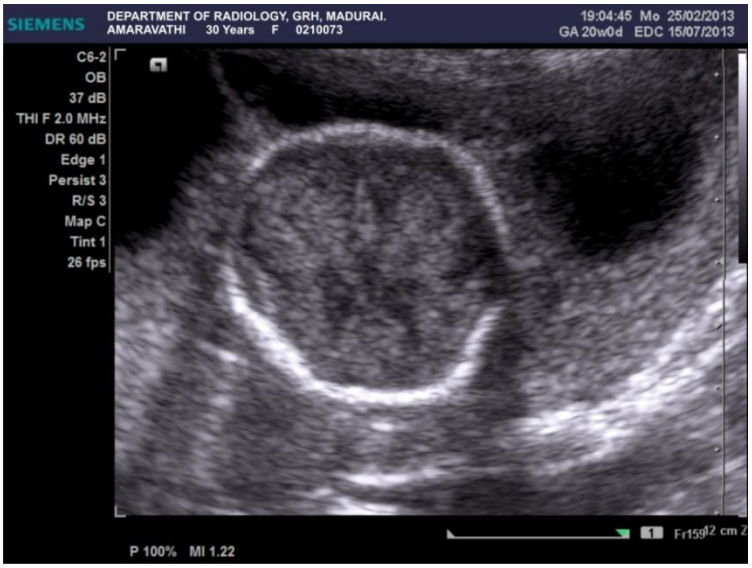




This distal obstruction of the urinary tract is more common in male fetuses, and the most common etiology is posterior urethral valves. Characteristically, there is dilatation of the bladder and proximal urethra.

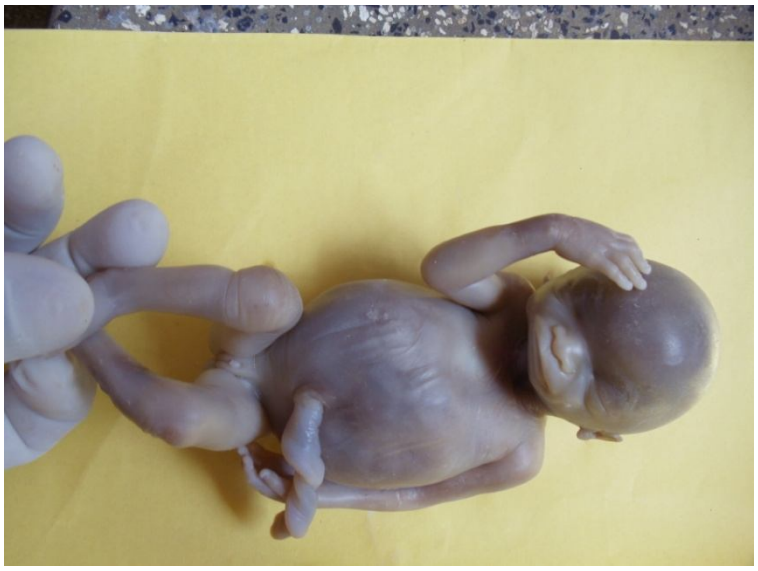
SCAN SHOWING CYSTIC HYGROMA

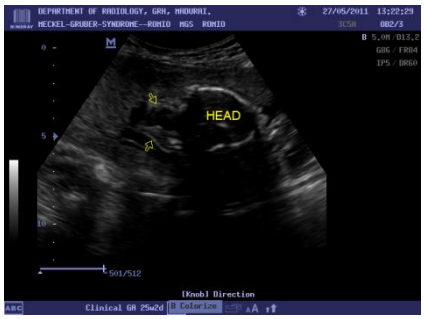


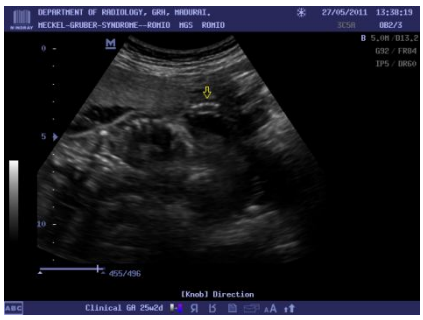


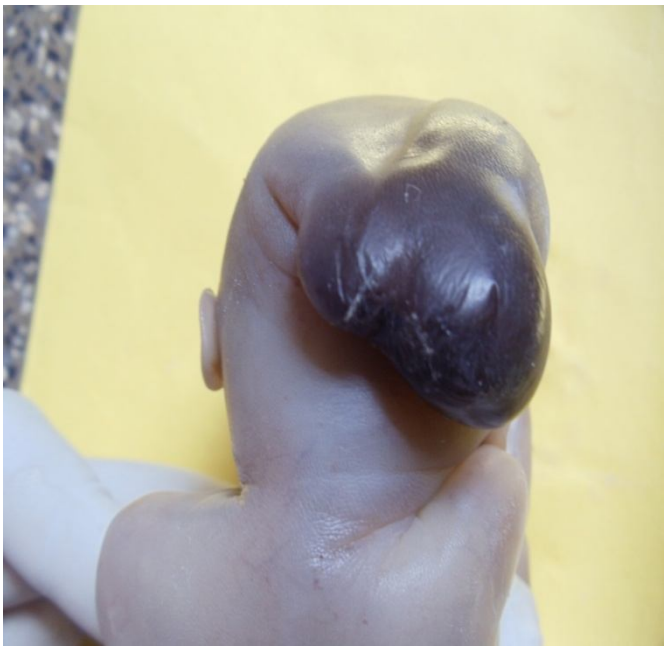
Cystic hygroma –this is a malformation of the lymphatic system in which fluid filled sacs extend from the posterior neck .They typically develop from lymphatic obstruction sequence,in which lymph from the head fails to drain in to the jugular lymphatic sacs .Enlarged thorasic duct can impinge on the developing heart associated with cardiac malformations .

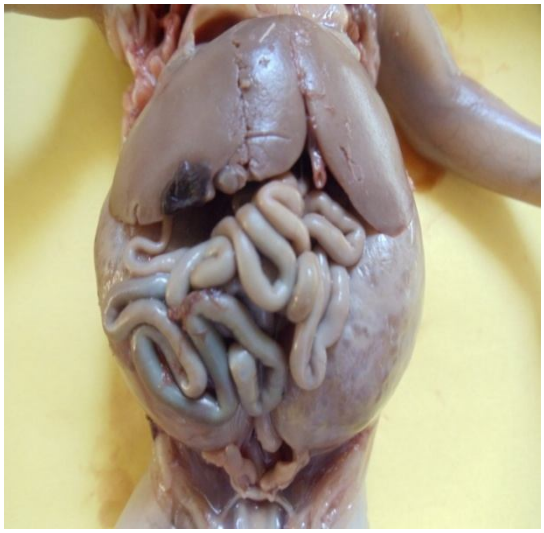
PHOTO AND SCAN SHOWING MECKEL GRUBER SYNDROME





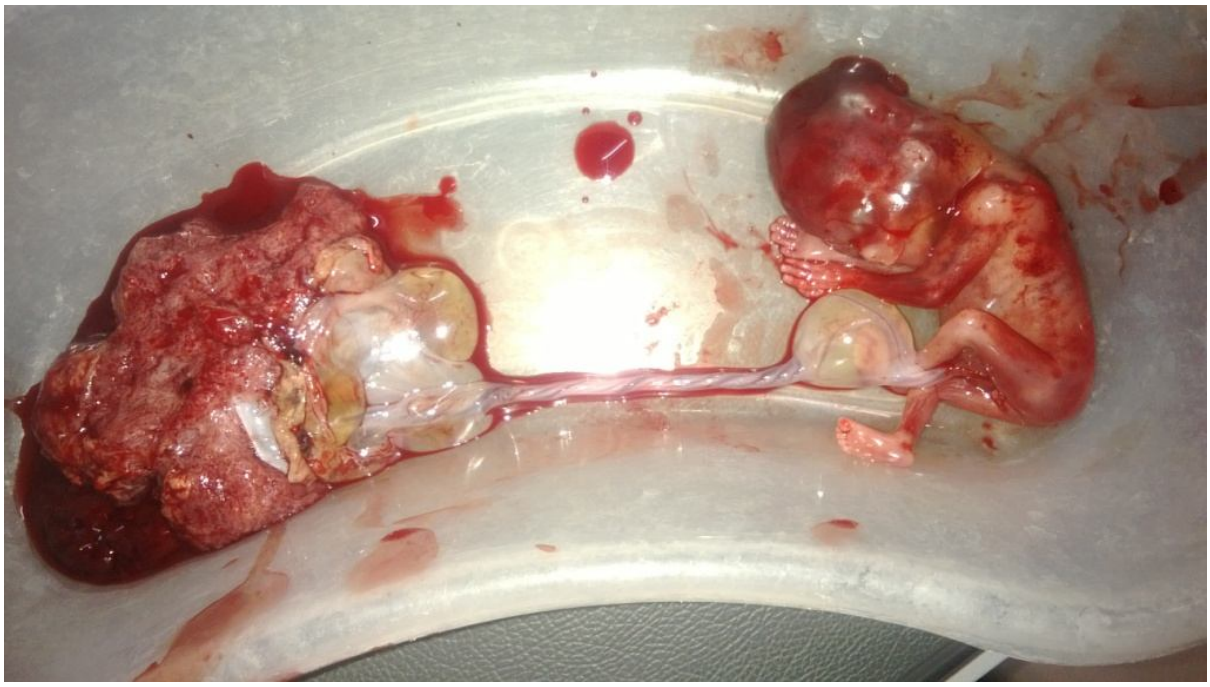


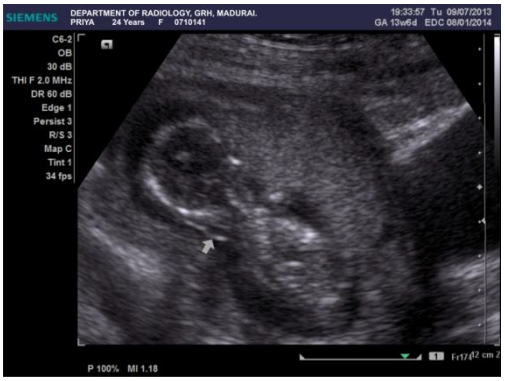


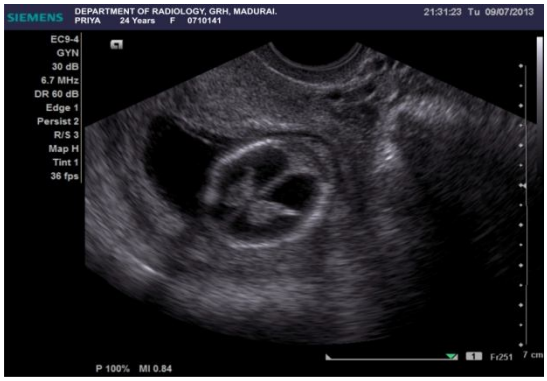


Encephalocele is the herniation of meninges and brain tissue through a cranial defect .Here MECKEL GRUBER SYNDROME with occipital encephalocele ,poly dactyly ,bilateral enlarged kidneys .

PHOTO SHOWING PARITEL ENCEPHALOCELE WITH HEMIVERTIBRAE



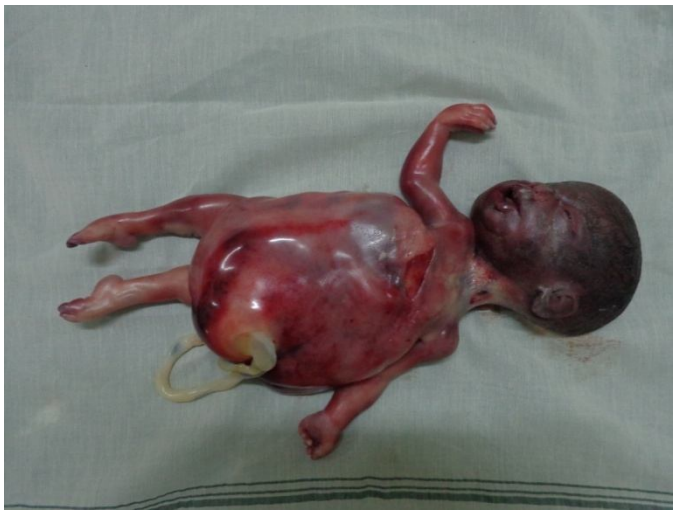


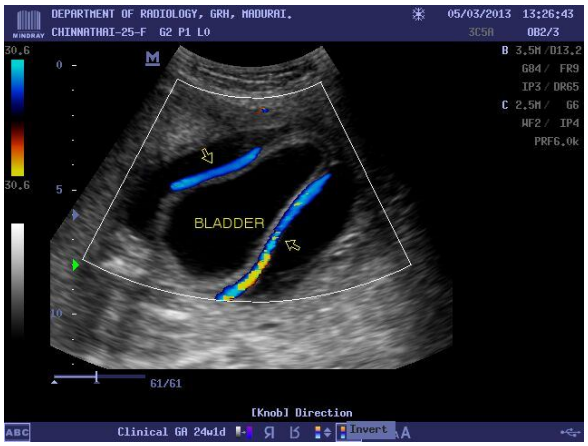


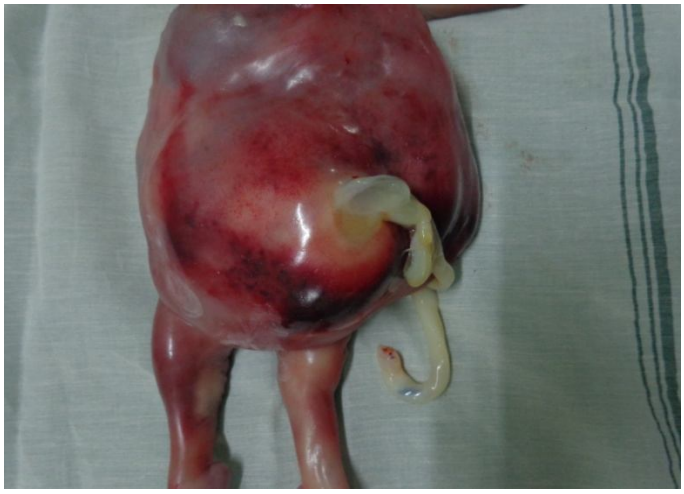
Open neural defect with Paritel Encephalocele With Hemivertibrae with umbilical cord cyst.

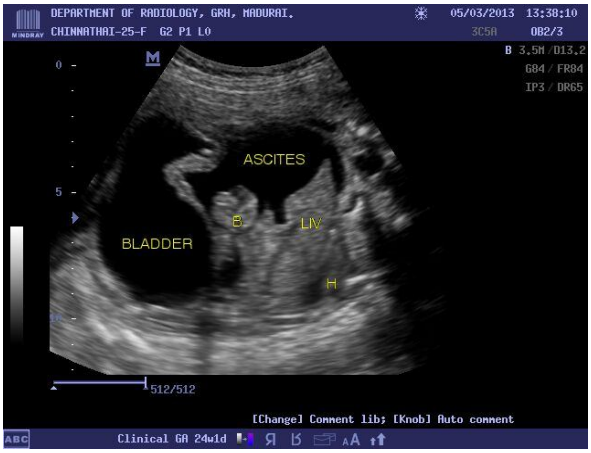
Incidence 1.4 – 2/100 pregnancies associated with aneuploidy, Trisomy 13, Trisomy 18, Triploidy.

PHOTO SHOWING URETHERAL ATRESIA WITH URINARY ACITES



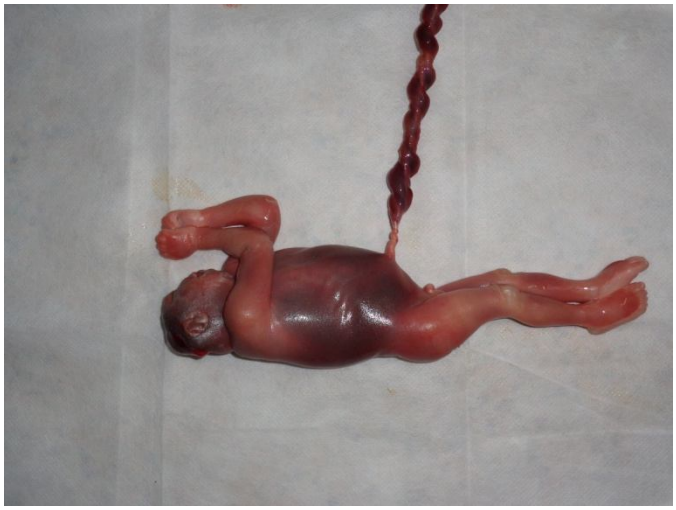


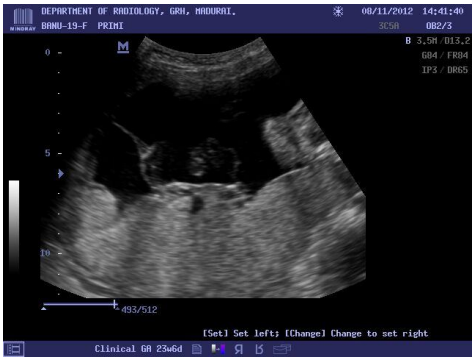


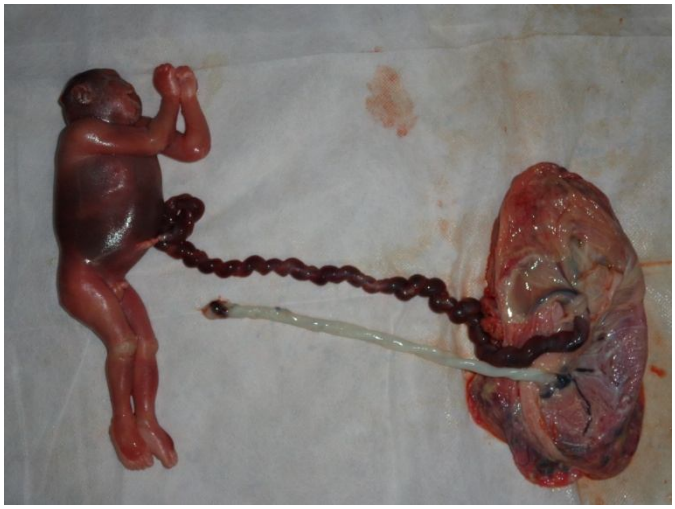


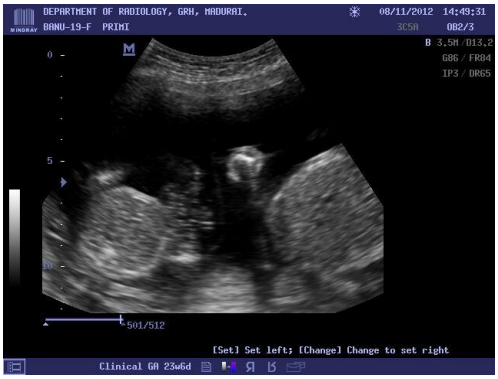
Dilated bladder with thickened wall in the setting of dilated proximal ureters, oligohydramnios, and enlarged multicystic kidneys. Follow up scans later in gestation revealed decompression of the fetal bladder and ureters, with accumulation of ascites thought to represent urinary ascites resulting from rupture of renal cysts.

PHOTO SHOWING TWINS WITH COARCTATION OF UMBILICAL CORD OF SINGLE FETUS WITH DEMISE









Coarctation is characterized by a localized narrowing of the cord with disappearance of the Wharton's jelly, thickening of the vascular walls, and narrowing of their lumens. Generally, torsion of the umbilical cord is present.

CONCLUSION

Prenatal diagnosis of congenital anomaly by USG is a noninvasive, readily acceptable, patient friendly mode of investigation for earlier diagnosis of fetal anomalies. It has got a high sensitivity of 93.7%, specificity of 98.8%.Positive predictive value of 93.75%, Negative predictive value of 98.% .The false positive rate is very low 1.19%.

By analyzing the risk factors mothers at high risk for anomalies can be subjected earlier even at 14 weeks for level II scan and early diagnosis of lethal congenital anomalies, and can be terminated, thereby decreasing the maternal morbidity, psychological upset and also decrease the workload of the health care provider.

Early detection of correctable anomalies compatible with life helps us to plan the mode of therapy and referral to higher centers for immediate ante partum, intrapartum and neonatal help, thereby decreasing the prenatal mortality and morbidity.

In case of neural tube defects we can decrease the incidence of recurrence by proper periconceptional folic acid supplementation therapy.

Consanguineous marriages to be avoided. General Nutritional Status to be improved.

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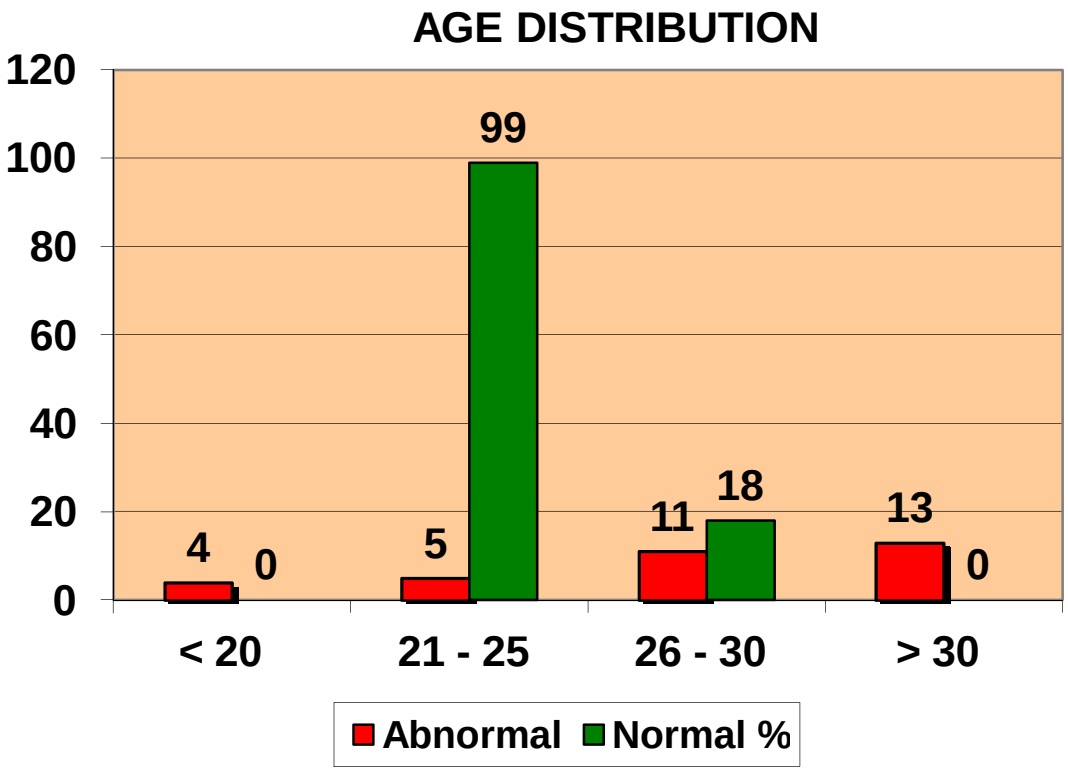
ABBREVIATIONS

N	-	Normal
CM	-	Consanguineous Marriage
NCM	-	Non Consanguineous Marriage
DM	-	Diabetes Mellitus
BA	-	Bronchial Asthma
EP	-	Epilepsy
HD	-	Heart Disease
M/S	-	Married Since
S	-	Sibling
H	-	Husband
P	-	Parent
FW	-	Fetal wastage
FA	-	Fetal Anomaly
C	-	Continuation
T	-	Termination
B	-	Booked
UB	-	Unbooked
NR	-	Non Reactive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18
13
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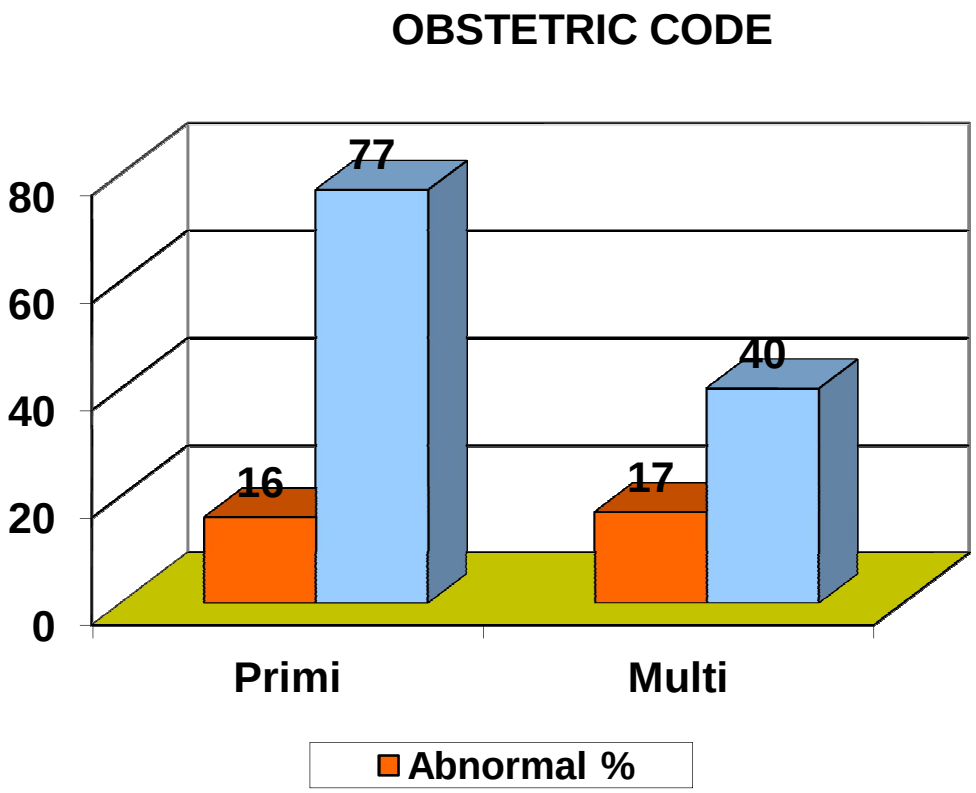
26 - 30
> 30
Abnormal
Normal %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Multi
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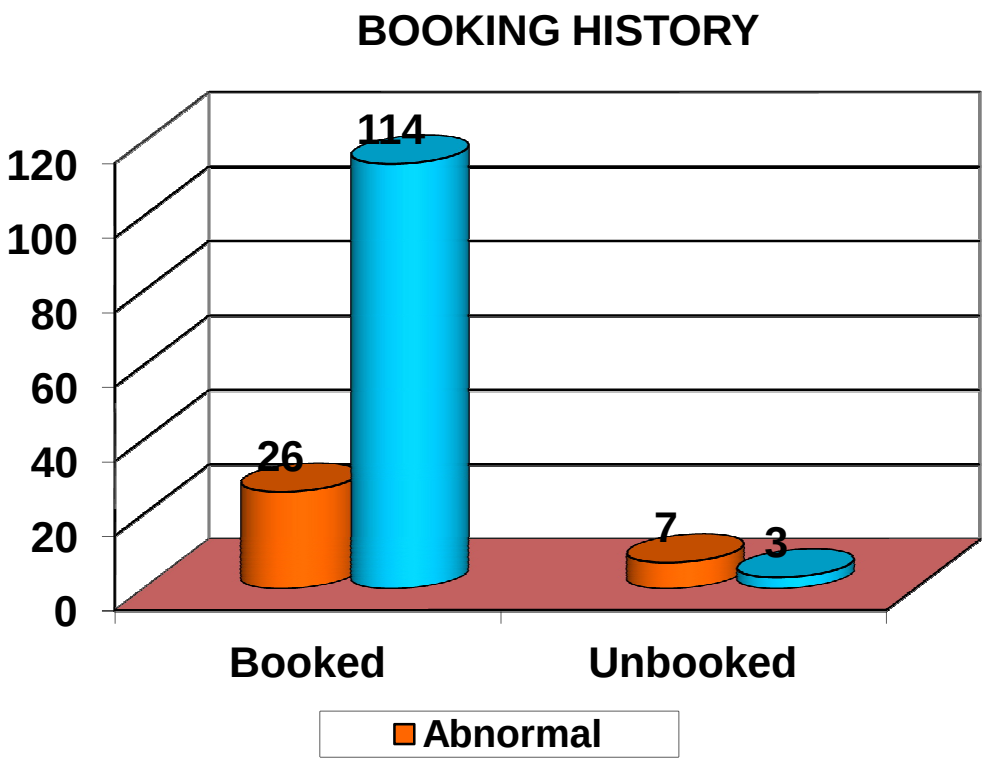
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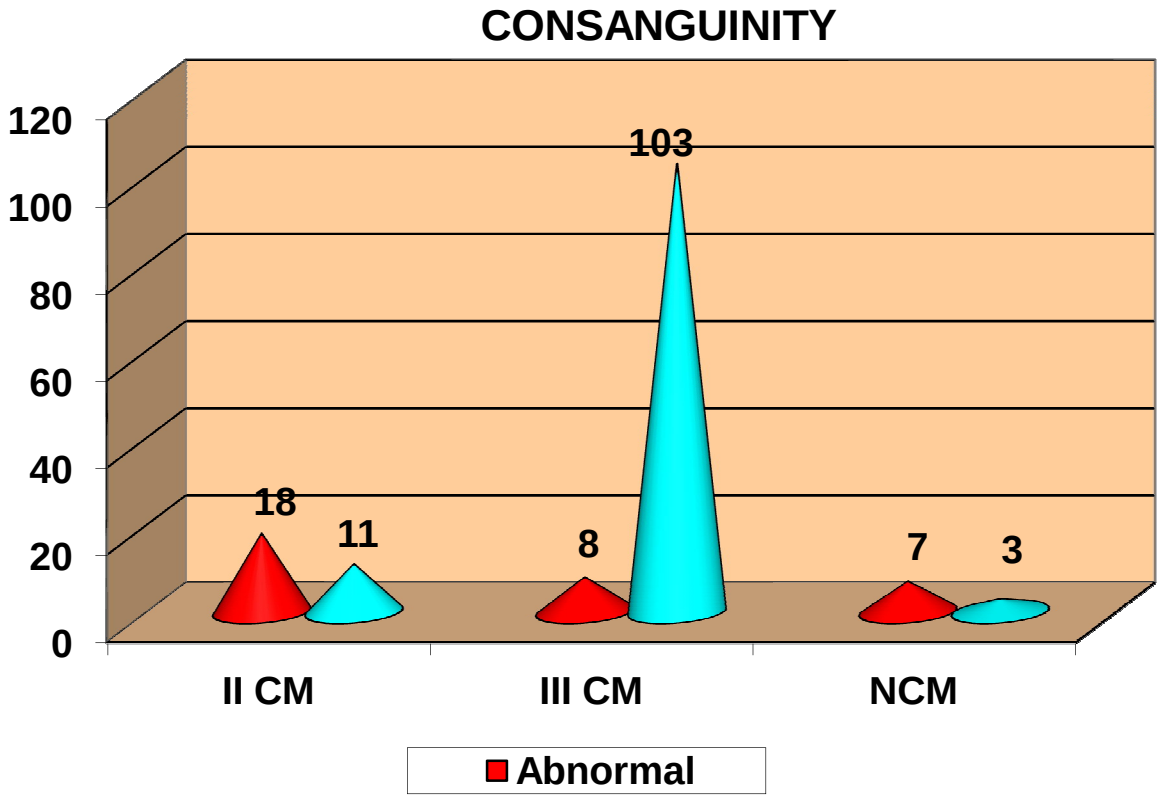
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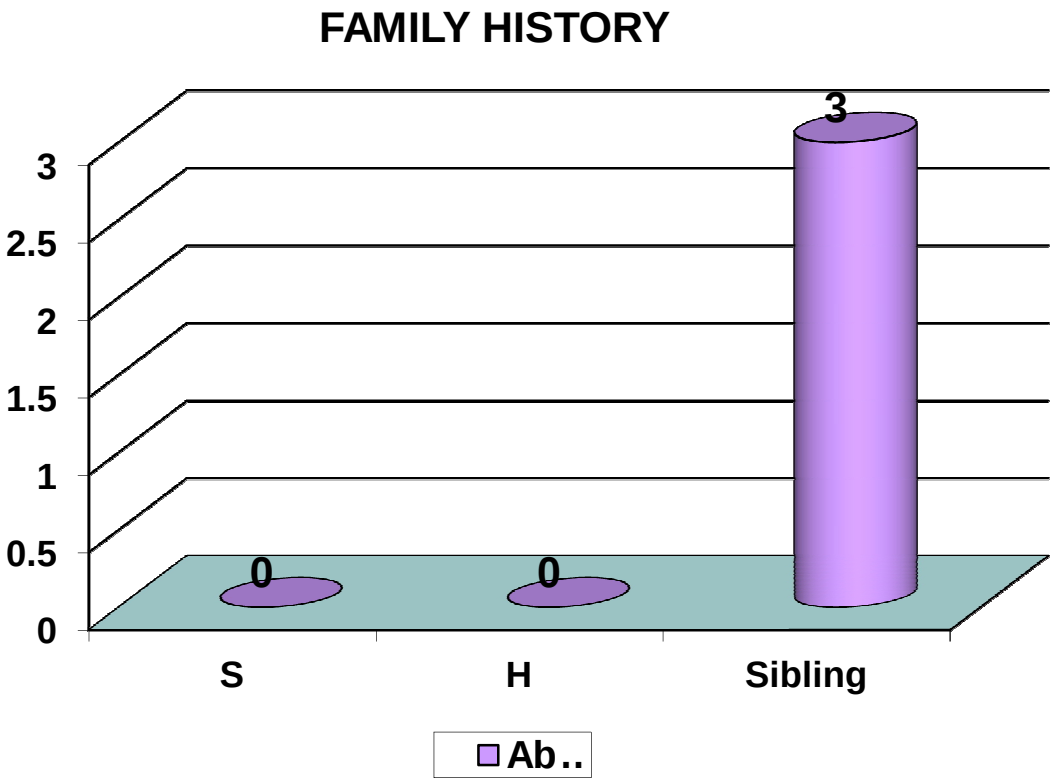
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Ab..

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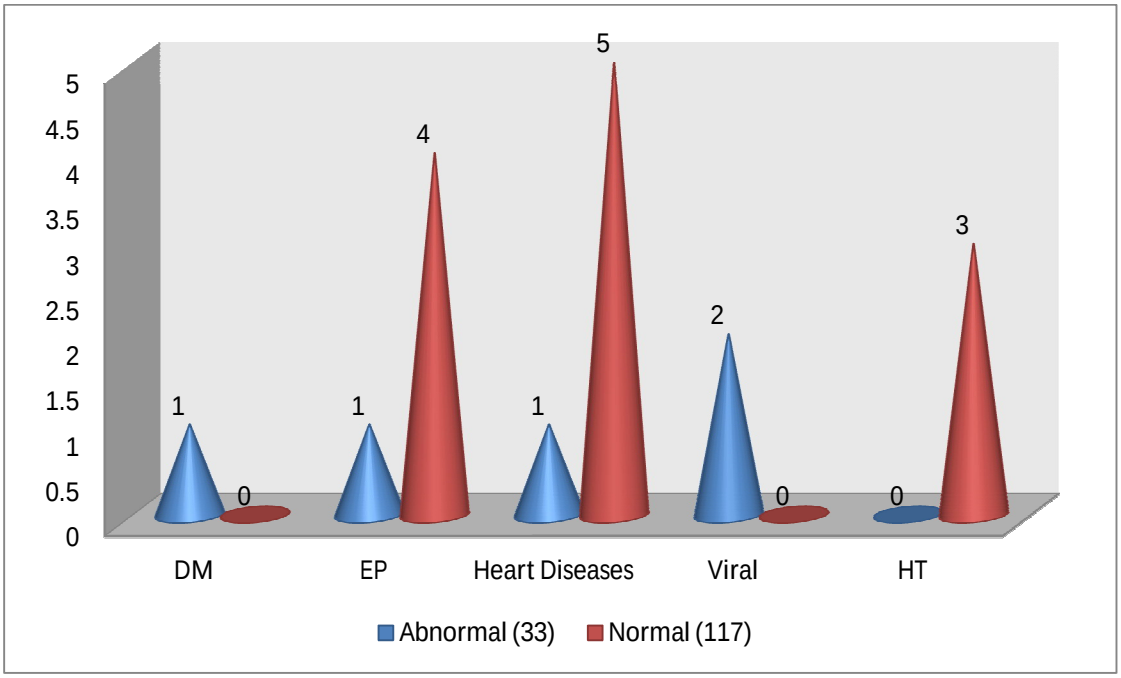

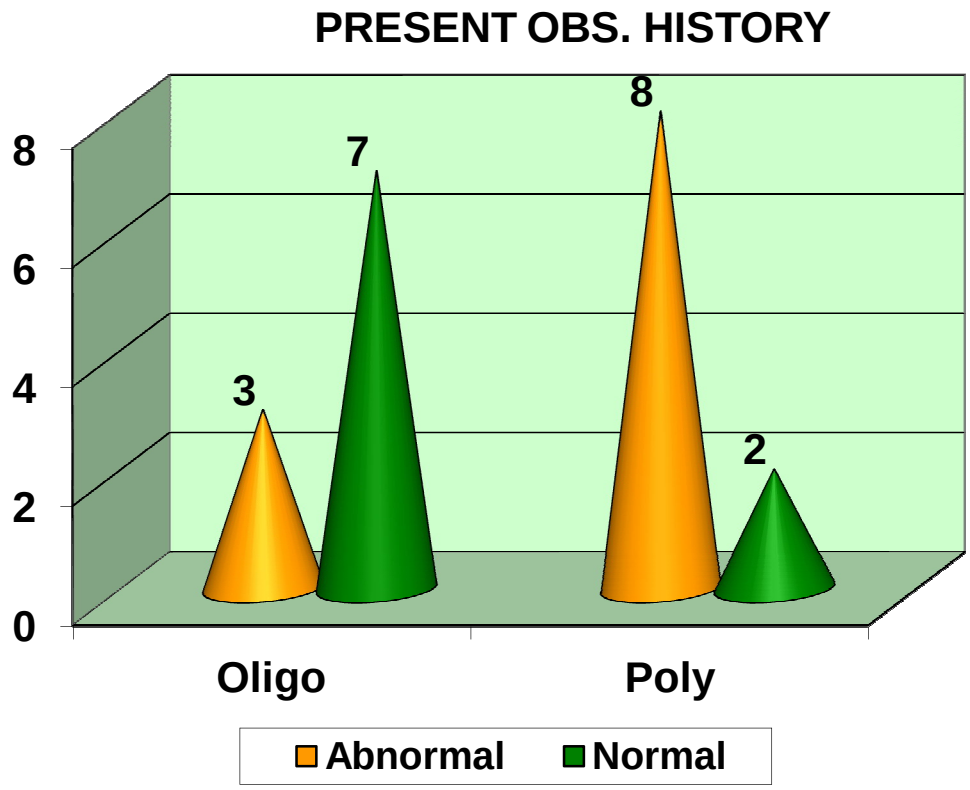
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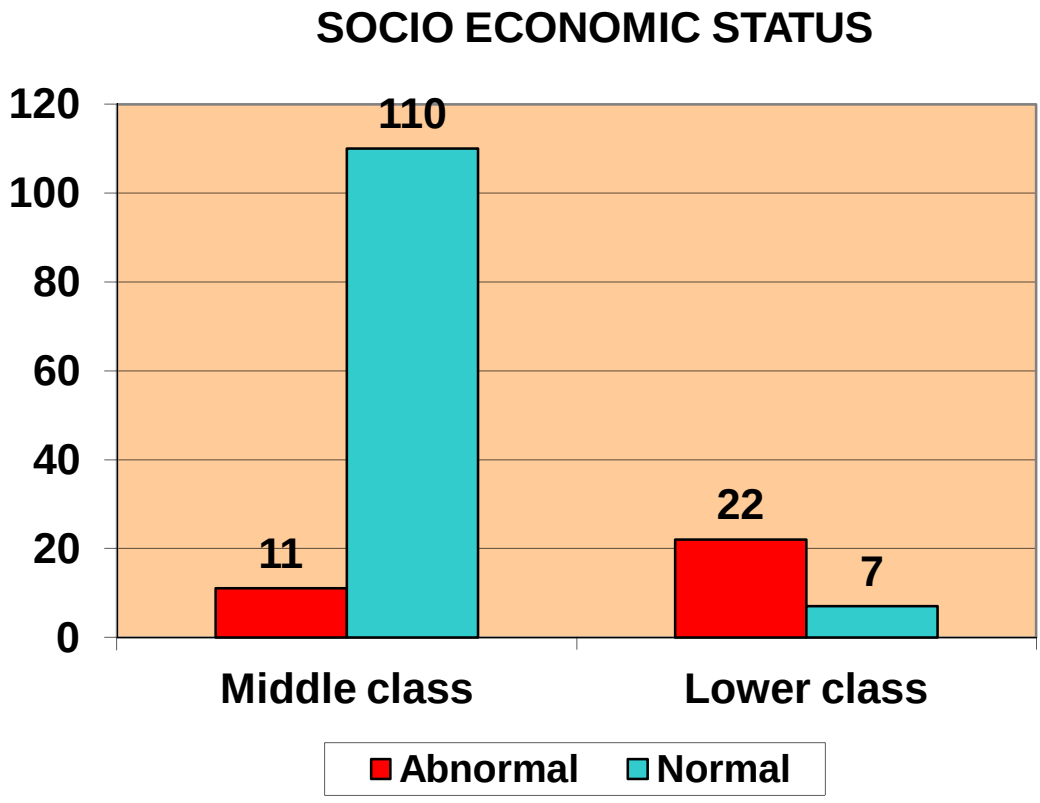
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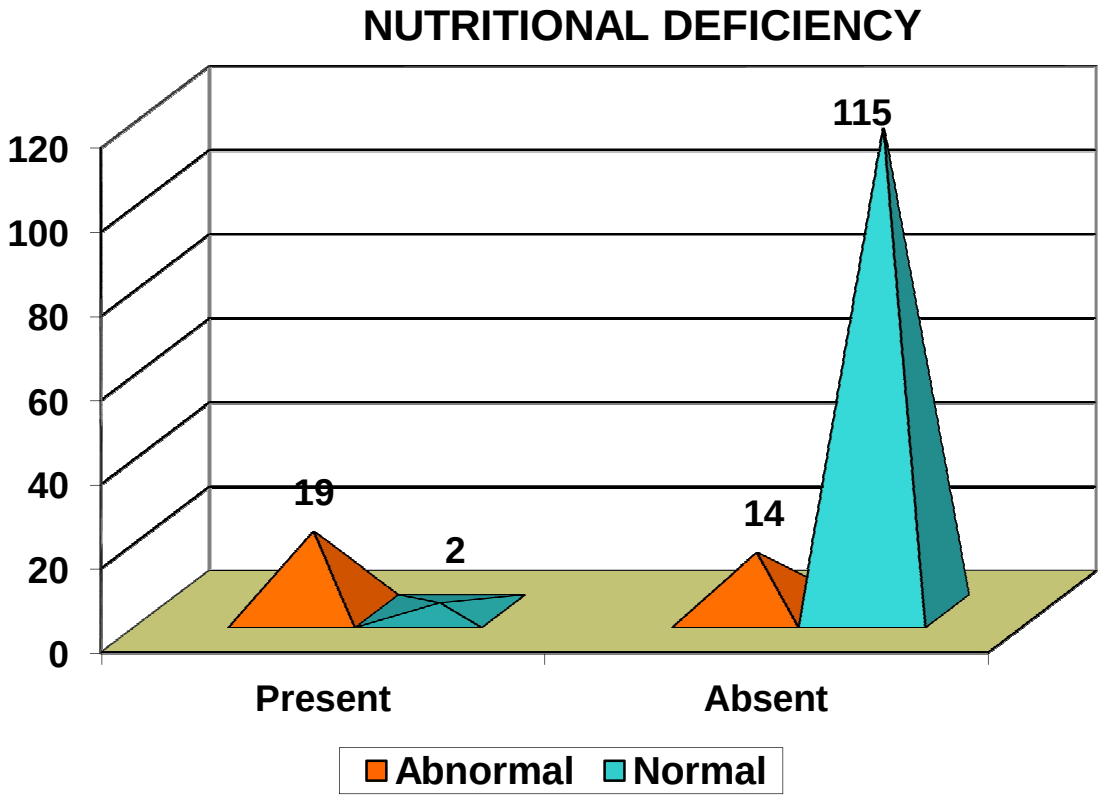
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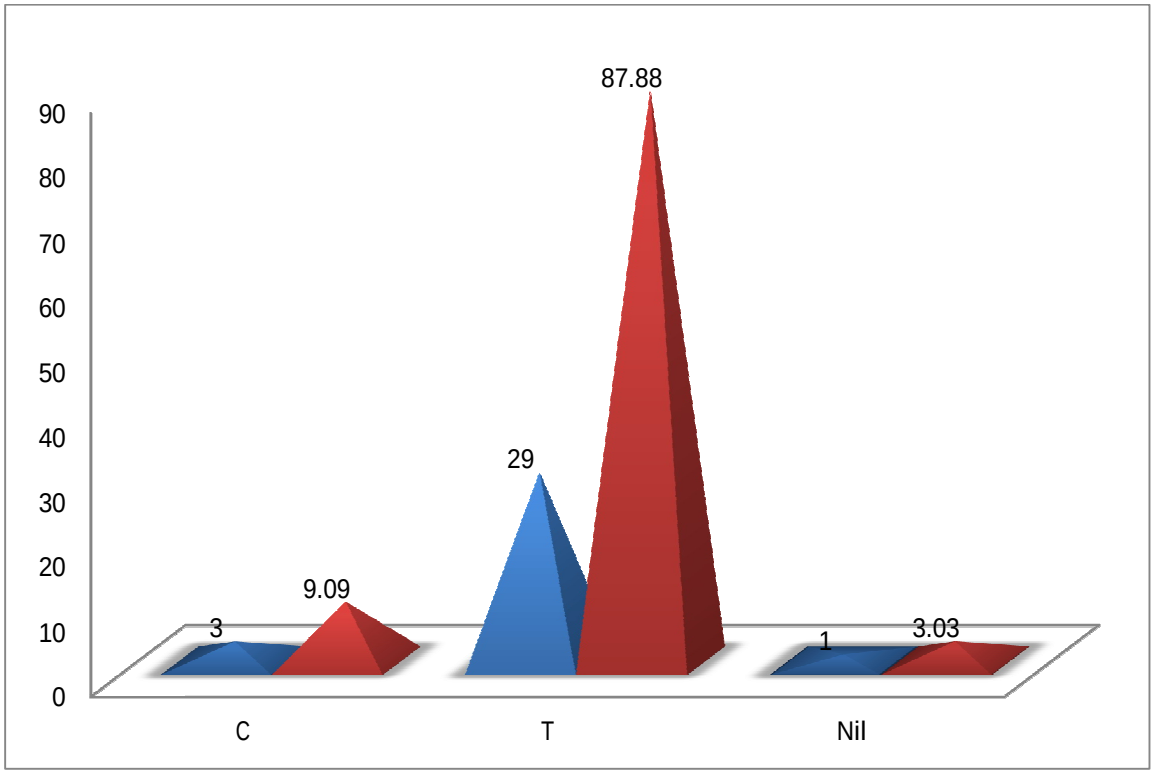
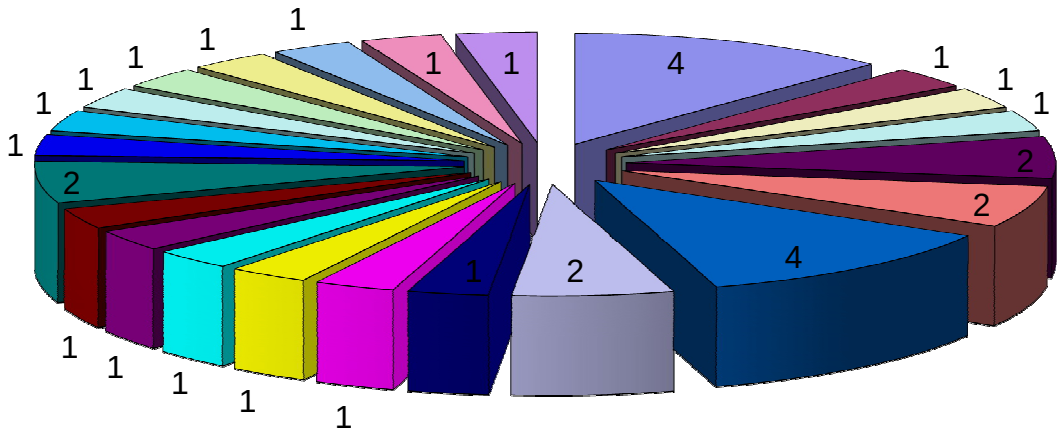
CONTINUATION / TERMINATION



USG FINDINGS



- Anencephaly
- Arnoldchiari malformation
- Bifrontal scalloping occipital flattening with Choroid Plexus Cyst
- Bilateral Renal Dysplasia
- Bladder Outlet Obstruction
- Congenital Diapragmatic hernia
- Conjoint Twins
- Craniochisis
- Cystic Hygroma
- Gastrochisis
- Aminiotic Band Syndrome
- Heterotaxy Syndrome
- Rocker Bottom Foot
- Iniencephaly
- Limb Body Wall Complex
- Meckel Gruber Syndrome
- Nonimmune hydrops
- paritel encephalocele with hemivertibrae
- Pulmonary &Tricuspid atresia with large VSD
- Mesomalic Type Of Skeletal Dysplasia
- TOF with Smallsize PA
- Twins With Coarctation Of Umbilical Cord Of Single Fetus
- Uretheral Atresia

USG Findings	No .of cases	Percentage
Normal	117	78
Abnormal	33	22
Total	150	

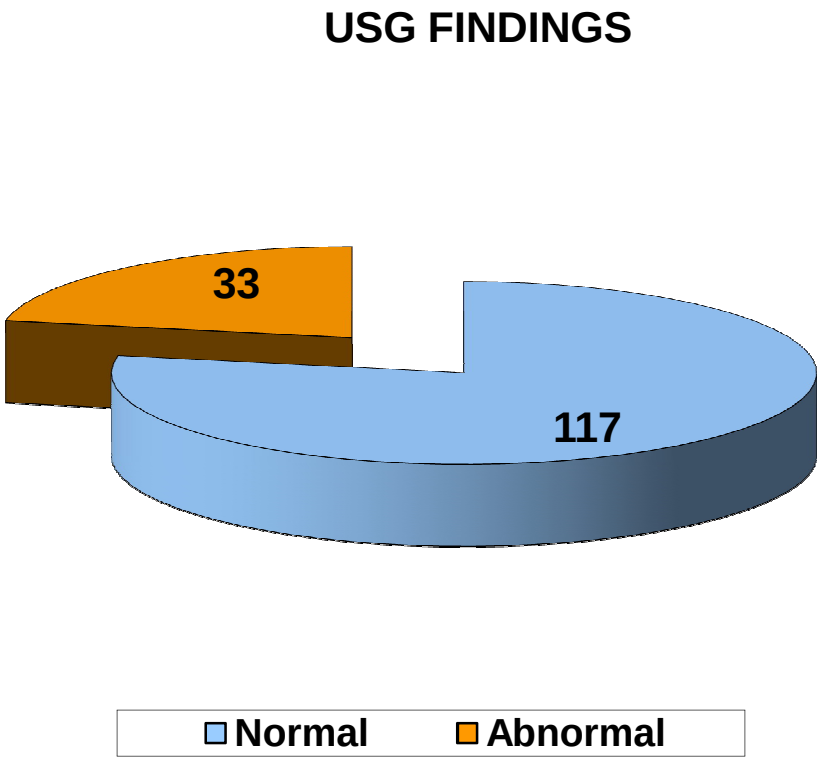


PHOTO SHOWING SACROCCYGEAL TERATOMA

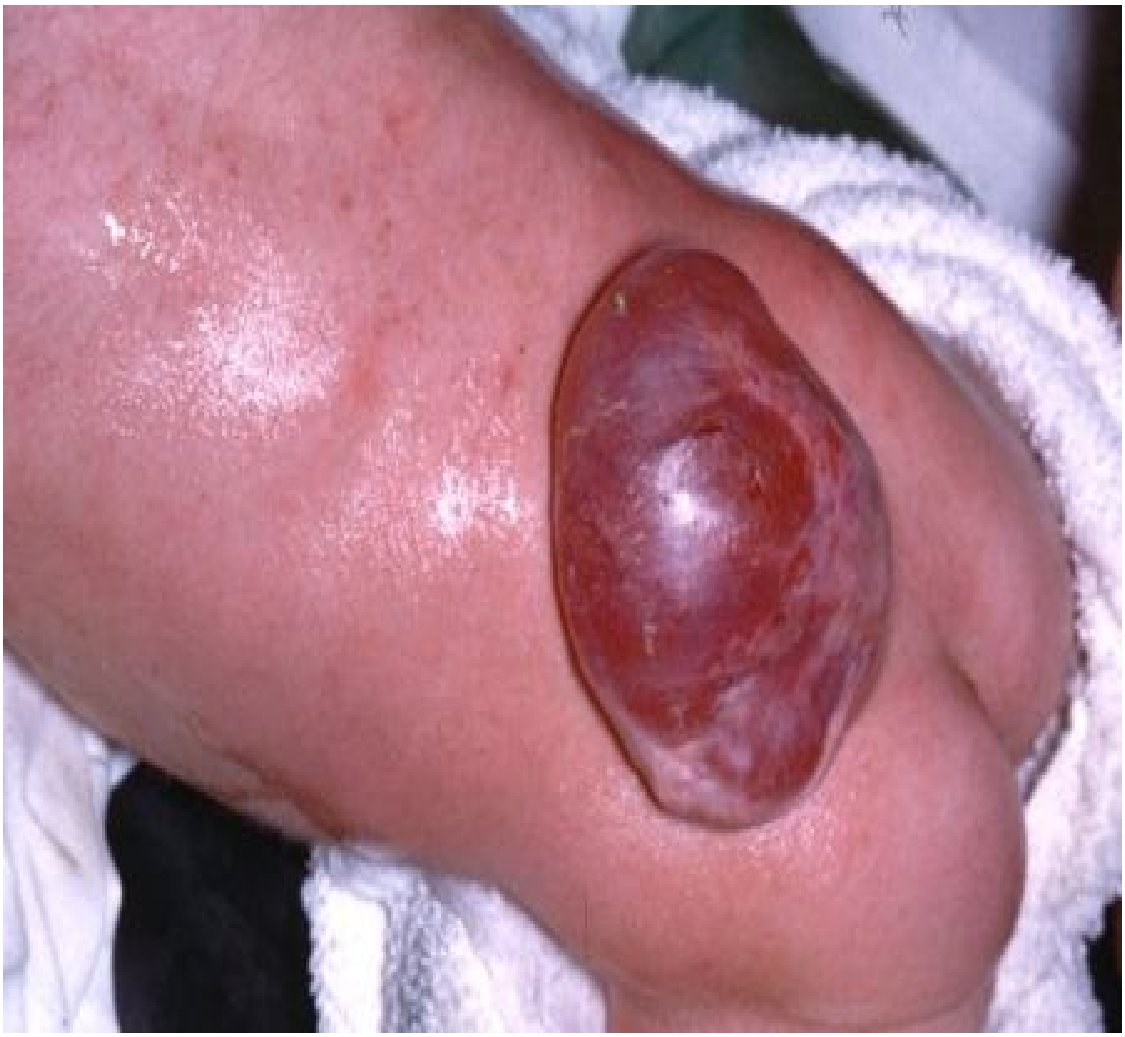


PHOTO SHOWING OMPHALOCOELE