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Study of fetal anomalies in second trimester of pregnancy in central India region

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Abstract

Congenital anomalies are single or multiple defects in development of fetus. Congenital anomalies are caused due to both genetic and environmental factors. Congenital anomalies are diagnosed either in intrauterine life or at birth.

Global incidence of congenital anomalies is 2-3%. Early detection of congenital anomalies during intrauterine life remains the key for further counselling and treatment. Diagnosis and treatment of congenital anomalies is a multidisciplinary assistance including obstetrician, genetic counsellor, embryologist, clinical geneticist and neonatologist.

Keyword: Congenital anomalies, neural tube defects, anencephaly, holoprosencephaly, spina bifida, pentalogy of Cantrell, duodenal atresia.

Introduction

People have always been interested in knowing how they were originated, developed, and were born, and why only some people develop abnormally. Ancient people, filled with curiosity, developed many answers to these questions.

For hundreds and thousands of years, the fetus has been an inaccessible patient surrounded by warm amniotic fluid and protected by uterus and anterior abdominal wall.

In ancient days, congenital abnormalities were only apparent at birth and were probably a cause of wonder and were considered to have a magical or symbolic importance. Indeed, many abnormalities have been incorporated into mythology and legend, such as the Mermaid based on Sirenomalia and Cyclops derived from Cyclopia associated with Holoprosencephaly.

The growth and development of human as a whole has always been matter of inquisitiveness for the mankind since the time immortal. Development of a human from cells has been supposed to be a study material in the civilized world for various reasons. The reasons may be

of prediction of better outcome, monitoring, developmental delay, anomalies as well as for legal purpose. In the days of single or two child norm every fetus in womb is supposed to be precious, imparting the need for evaluation of its development.

In the early 20th century major anomalies such as severe hydrocephalus could be suspected from clinical examination and confirmed by plain radiology. However, it was not until the advent of obstetric ultrasound that the full range of fetal disease could be diagnosed and this came out in the concept of the "Fetus as a patient."

Routine obstetric scanning in the second trimester was introduced into the United Kingdom in the early 1980s. In 1984, a report of a working party of the Royal College of Obstetricians and Gynecologists recommended that this scan should be performed between 18 to 20 weeks of gestation. 2 years later, a Joint Study Group suggested that all normal pregnant women should be offered a detailed anomaly scan by 20 weeks of gestation.

Other important revolutions in the field of ultrasonography are Colour Doppler, 3D and 4D ultrasonography, Colour Doppler and power colour flow imaging.

Ultrasound scans may provide important information about fetal anatomy. Parents should be aware of the goals and limitations of this type of examination. In the event of lethal or severe malformations, they will face a choice between proceeding with the pregnancy or considering termination. Ultrasound examination may make it possible to plan birth in a tertiary center ensuring adequate neonatal management. In some cases prenatal ultrasound may result in an unnecessary burden of anxiety, but it should be stressed that in the vast majority of cases its reassuring effect is of very significant benefit to the parents.

In this respect, an ultimate aim has been achieved: Once we have the fetal patient, it is inevitable there should be fetal therapy.

Material & methods

The project "The study of Fetal Anomalies In second Trimester of Pregnancy By Ultrasonography In Central India Region" was carried out in our medical college and hospital, over the period of 2 years.

The study included 1000 pregnant women who were selected from the antenatal care (ANC) patients in second trimester i.e., between 12 and 24 weeks of gestation attending ANC clinic for ultrasonographic screening at our medical college and hospital. The study subjects included rural, urban and migrated population from most part of the Central India.

The subjects selected were from the age group of 18 to 40 years. All the subjects had sound knowledge about their menstrual dates. The subjects were given prior appointment in the morning hours and screened under the guidance of only one experienced sonologist throughout the study.

The following procedure was adopted to collect the data for the study. The study patients were informed regarding the nature and purpose of the study and written consent was taken from the patients in the presence of an impartial witness.

1. Particulars of the subjects selected for the study with special reference to their menstrual and obstetric history, drug history was recorded in the proforma given below.
2. Ultrasound screening examination was performed on the subjects with full urinary bladder.
3. The equipment used was e saote My Lab 50 X vision which was routinely used in the obstetric practice.

4. The subjects were positioned on the ultrasound screening table in supine position with their abdomen exposed. Then a mineral oil gelly, 'Sonogel' was applied all over the surface so as to ensure an airless contact between the tissue and the transducer probe.

5. Transducer probe was placed in the longitudinal direction and moved all over the surface of the abdomen. The fetal position was then assessed.

6. Following particulars were noted

- Number of gestations.
- Presentation of the fetus at the time of examination.
- Approximate gestational age of the fetus in weeks was obtained with the help of fetal parameters like fetal Biparietal Diameter (BPD), fetal Head Circumference (HC), fetal Abdominal Circumference (AC); fetal Femur Length (FL) as per the recommendations of the American Institute of Ultrasound in Medicine.
- The umbilical cord was identified and traced till its insertion in the placenta. First longitudinal scanning and then transverse scanning was done. Thus, the position and extent of placenta was determined and its relativity from cervix was noted.
- For assessment of amniotic fluid, amniotic fluid Index (AFI) was determined during ultrasound examination as per procedure given below:
- Patients were given supine position.
- A linear transducer was used.
- Maternal uterus was divided into four quadrants by a sagittal midline vertical plane and an arbitrary transverse line approximately halfway between the symphysis pubis and upper edge of the uterine fundus.
- The transducer was kept parallel to the maternal sagittal plane and perpendicular to the maternal coronal plane throughout examination.
- The deepest unobstructed and clear pocket of amniotic fluid was visualized and the image was obtained. The ultrasound caliper's were manipulated to measure the pocket in a strictly vertical direction.
- The process was repeated in each of the four quadrants and the pocket measurements were summed = AFI
- If the AFI was <8cm, the evaluations in all four quadrants were performed three times and average values were taken.
- Polyhydramnios was diagnosed if AFI > 24cm or a single pocket of fluid at least 8 cm in depth.
- Oligohydramnios was diagnosed if AFI ≤ 5cm.
- Fetus was screened for the structural anomalies of the head, face, neck, thorax, abdomen, limbs, spine and genitourinary system.
- The information collected by the above procedure was recorded in the following proforma.

Results

The study was conducted on 1000 pregnant women who were attending antenatal clinics for routine examination and ultrasonographic screening in between 12 to 24 weeks of gestation at our Medical College and Hospital. All the subjects had sound knowledge about their menstrual dates and previous menstrual cycles.

The parameters considered in our study were maternal age, gravida, gestational age, drug history, fetal anomalies, types of fetal anomalies, polyhydramnios or oligohydramnios associated with fetal anomalies.

Table 1: Shows Maternal age wise distribution of study subjects.

Maternal age (Years)	No. of subjects N=1000	%
<20	44	4.4
20-24	593	59.3

25-29	260	26.0
>30	103	10.3

In this study the maternal age ranged from 18 to 40 years. Mean maternal age in years = 24.08±3.61

Table 2: Shows Gravida wise distribution of study subjects.

Gravida	No. Of subjects N=1000	%
1	548	54.8
2	356	35.6
3	81	8.1
4	11	1.1
>4	4	0.4

Study subjects were ranged from gravid 1 to gravid 7. Mean gravida = 1.57±0.74

Table 3: Shows week wise distribution of Gestational Age.

Gestational Age (Week)	No. of subjects N=1000	%
12-16	100	10.0
17-21	391	39.1
22-24	509	50.9

In this study gestational age ranged from 12 to 24 weeks. Mean Gestational Age = 20.82±2.64

Table 4: Shows incidence of fetal anomalies.

USG findings	No. of subjects N=1000	%
Fetal anomalies	31	3.1

In this study 1000 pregnant women were studied. Out of those, fetal anomalies were observed in 31 pregnancies. Incidence of fetal anomalies was calculated as 3.1 %.

Table 5: Shows system wise distribution and incidence of fetal anomalies.

System	No of subjects	Incidence/1000
Neural Tube Defects	10	10
Facial cleft	1	1
Cystic hygroma	1	1
Cardiovascular defects	3	3
Congenital diaphragmatic hernia	1	1
Abdominal wall defect.	1	1
Gastro-intestinal Defect	1	1
Simple Ovarian cyst	3	3
Urinary system defects	3	3
Musculoskeletal disorder	4	4
Hydrops fetalis	2	2
Syndrome- Pentalogy of Cantrell	1	1
Total	31	31

Neural tube defects were observed in 10 cases. Neural tube defects included Anencephaly (n=3), Holoprosencephaly (n=2), Aqueductal stenosis (n=2), Hydrocephalus (n=1), Occipital encephalocele (n=1), Dandy Walker cyst (n=1). Incidence of neural tube defects was calculated as 10 per 1000 pregnancies.

Facial cleft was observed in 1 case.

It was cleft lip on right side. Incidence of facial cleft was found 1 per 1000 pregnancies.

Cystic hygroma was observed in 1 case giving an incidence of 1 per 1000 pregnancies.

Fetal cardio-vascular anomalies were found in 3 cases. All 3 cases of cardiovascular anomalies were ventricular wall defects. Incidence of cardiovascular anomalies was calculated as 3 per 1000 pregnancies.

Fetal diaphragmatic hernia of Bochdalek type was observed in 1 case. Incidence of fetal diaphragmatic hernia was found 1 per 1000 pregnancies.

Fetal anterior abdominal wall defect showing Gastroschiasis was observed in 1 case giving an incidence of 1 per 1000 pregnancies.

Gastro-intestinal defect was seen in 1 case. It was Duodenal atresia.

Incidence of gastro-intestinal tract defect was found as 1 per 1000 pregnancies.

Simple ovarian cyst was seen in 3 cases. Those were one simple ovarian cyst on left side and 2 cases of simple ovarian cysts on right side. Incidence was calculated as 3 per 1000 pregnancies.

Urinary system defects were seen in 3 cases. All the three defects were fetal polycystic kidney disease with hydronephrosis. Incidence of urinary system defects was 3 per 1000 pregnancies.

Musculoskeletal disorders were seen in 4 cases. Disorders included one case of congenital talipus equinovarus and 3 cases of short limb dwarfism. Incidence of musculoskeletal disorders was 4 per 1000 pregnancies.

Hydrops fetalis was seen in 2 cases giving an incidence of 2 per 1000 pregnancies.

Multiple fetal anomalies were seen in 1 case showing single anterior abdominal wall defect with a covering membrane, sternal cleft with ectopia cordis, kyphoscoliosis, bilateral pleural effusion with mild generalized subcutaneous edema, suggested pentalogy of Cantrell syndrome.

Graph 1 shows system wise distribution and incidence of fetal anomalies.

Table 6: Shows percent wise distribution of fetal anomalies.

System	N=31	%
Neural Tube Defects	10	32.3
Facial cleft	1	3.2
Cystic hygroma	1	3.2
Cardiovascular defects	3	9.7
Congenital diaphragmatic hernia	1	3.2
Abdominal wall defect	1	3.2
Gastro-intestinal Defect	1	3.2
Simple Ovarian cyst	3	9.7
Urinary system defects	3	9.7
Musculoskeletal defects	4	12.9
Hydrops fetalis	2	6.5
Syndrome- Pentalogy of Cantrell	1	3.2

- Neural tube defects were observed in 32.3% of fetal anomalies.
- Facial cleft was observed in 3.2% of fetal anomalies.
- Cystic hygroma was observed in 3.2% of fetal anomalies.
- Cardiovascular defects were observed in 9.7% of fetal anomalies.
- Congenital diaphragmatic hernia was observed in 3.2% of fetal anomalies.
- Anterior abdominal wall defect was calculated as 3.2% of fetal anomalies.
- Gastrointestinal defects were seen in 3.2% of fetal anomalies.
- Simple ovarian cysts were observed in 9.7% of fetal anomalies.
- Urinary system showed 9.7% of fetal anomalies
- Musculoskeletal disorders were observed in 12.9% of fetal anomalies.
- Hydrops fetalis were seen in 6.5% of fetal anomalies.
- Multiple anomalies characterized as Pentalogy of Cantrell was observed in 3.2% of fetal anomalies.
- Most commonly observed anomalies were Neural tube defects with incidence of 32.3% of fetal anomalies.

Graph 2 shows percent wise distribution of fetal anomalies.

Discussion

Ultrasonography is an advanced tool for rapid obstetric imaging with high sensitivity without any adverse effects. Ultra sound scan is currently considered to be a

safe, non - invasive, accurate and cost-effective investigation in the fetus. It has progressively become an indispensable obstetric tool and plays an important role in the care of every pregnant woman.

The main use of ultrasonography is in diagnosis and confirmation of early pregnancy, vaginal bleeding in early pregnancy, determination of gestational age and assessment of fetal size, diagnosis of fetal malformation, placental localization, multiple pregnancies and polyhydramnios or oligohydramnios.

Table **7:** Incidence of fetal anomalies

Investigators	Incidence of fetal anomalies in %
Singh M (1980)[51]	2.7
Himmetogluo (1996)[20]	1.11
Dillon E (1997)[15]	2
Norchi H (1997)[43]	3.71
Van Dorsten JP (1998)[57]	2.3
Levi S (1998)[34]	2.5
Magriples U (1998)[38]	3.1
Long G (1998)[35]	2
Singh S (2006)[52]	1.14
Lalor J (2007)[32]	2
Akinola RA (2008)[2]	0.73
Present study	3.1

Singh M (1980)[51] studied 7,274 infants and 170 (2.7 per cent) subjects were diagnosed to have 241 major congenital malformations. The incidence of major malformations among preterm infants was almost twice (5.3%). Himmetoglu O (1996) [20] conducted study on 9160 neonates the overall congenital anomaly incidence was 1.11%.

Dillon E (1997) [15] found fetal abnormalities in 2% of cases. Narchi H (1997) [43]represented an incidence of 37.1 anomalies per 1000 total births. Similar study was carried out by Van Dorsten JP (1998) [57]in South Carolina and diagnosed 2.3% of fetal abnormalities by ultrasonography. According to Levi S (1998)[34] the prevalence of fetal malformations was as high as 6.5% but only 2.5% of those were potentially life threatening or representing major cosmetic defects. Magriples U (1998)[38] during their study found out 3.1% of incidence of fetal anomalies. In the study carried out by Long G (1998)[35] 2% of all the babies were born with the major structural abnormalities. According to Singh S (2006)[52] study the major congenital malformations were 1.14%.

In Ireland Lalor J (2007)[32] carried out ultrasound screening for fetal abnormalities. The incidence of fetal anomalies was 2%. Akinola RA et al (2008)[2] conducted an audit of congenital fetal anomalies as seen on ultrasound scan. Congenital fetal anomalies were detected in 0.73% of cases.

In present study 31 fetal anomalies are detected out of 1000 cases. The incidence of fetal anomalies is calculated as 3.1%. This finding is comparable with the studies conducted by Singh M (1980)[51], Dillon E (1997)[15], Narchi H (1997)[43], Van Dorsten JP (1998)[57], Magriples U (1998)[38] and Lalor J (2007)[32]. Slight variations are considered negligible from statistical point of view or it may be due to difference in ethnicity.

Table 8: Neural Tube Defects

Investigators	Incidence of fetal anomalies in %
Kulkarni ML (1989)[30]	11.4
Mahadevan B (2005)[39]	5.7
Safdar OY (2007)[48]	1.3
Khattak ST (2008)[28]	13.9
Al – Ain Zr (2010)[3]	3.3
Present study	10

Neural tube defects were studied by Kulkarni ML (1989)[30], Mahadevan B (2005)[39], Safdar OY (2007)[48], Khattak ST (2008)[28]and Al-Ain ZR (2010)[3].

Kulkarni ML (1989)[30] during the study an incidence of neural tube defects was 11.4/1000 cases. Similar study was carried out by Mahadevan B (2005)[39]. The incidence of neural tube defects was 5.7 per 1000 cases. During the study of Safdar OY (2007)[48]incidence of Neural Tube Defects was 1.3/1000 live births.

Khattak ST (2008)[28] found 46 Neural Tube Defects from 3310 subjects. Incidence of Neural Tube Defects in their study was 13.90 per 1000 deliveries. During the study conducted by Al-Ain ZR (2010)[3], 33 infants were delivered with Neural Tube Defects giving an incidence of 3.3/1000 births.

In the present study the incidence of neural tube defects is 10 per 1000 cases. Neural tube defects are 32.2% of anomalies found in our study.

These findings are comparable with Kulkarni ML (1989)[30]and Khattak ST (2008)[28]. The previous studies with lower incidence of Neural Tube defects were carried out in subjects with prior administration of folic acid.

Table 9: Facial Clefts

Investigators	Incidence /1000
Wayne C (2002)[59]	1.3
Present study	1

Orofacial clefts were studied by Wayne C (2002)[59]. During their study, the overall incidence was 1.3 in 1000 cases. During the study conducted by Ballet CR (2004)[11] 29 orofacial clefts were studied. The cleft was bilateral in 9 cases (31%), 6 of which involved the lip, alveolus, hard and soft palate, one cleft lip and alveolus, one isolated cleft lip and one Oro-ocular cleft. The cleft was unilateral in 18 cases (61%).

In the present study facial cleft is observed in 1 per 1000 cases. Incidence of facial clefts is observed as 3.2% of anomalies. It is cleft lip on right side. These findings are comparable with the study of Wayne C (2002)[59].

Cystic Hygroma

Oak SN (1992)[44]and Amin U (2007)[4], reported one case each of cystic hygroma. Oak SN (1992)[44] diagnosed a case of posterior midline cervical cystic hygroma in 18 weeks fetus prenatally. It was a multi septate cystic structure posterior to fetal head and neck without vertebral column defect. Similarly, Amin U (2007)[4] studied a cystic structure extending postero-laterally on both sides of fetal head and neck. They diagnosed it as cystic hygroma.

In the present study a cystic lesion is observed in the neck. No other congenital anomaly detected. The case is diagnosed as cystic hygroma. These findings are comparable with Oak SN (1992)[44] and Amin U (2007)[4]. The incidence of cystic hygroma is calculated as 1 per 1000 pregnancies. Cystic hygroma comprised 3.2% of anomalies.

Table 10: Cardiovascular System anomalies

Investigators	Incidence /1000
Narchi H (1997)[43]	3.3
Hoffman JIE (2002)[21]	4
Barbosa JM (2002)[10]	8
Archer man RJ (2007)[1]	2
Sipek A (2010)[53]	19.9
Present Study	3

Fetal cardiovascular system abnormalities were studied by Narchi H (1997)[43], Hoffman JIE (2002)[21], Barbosa JM (2002)[10], Hui L (2005)[24], Archer man RJ (2007)[1]andSipek A (2010)[53]. During study of Narchi H (1997)[43], 674 congenital malformations were diagnosed from 18,146 subjects. There were 60 subjects with congenital heart disease (3.3 per 1000 subjects). The

most common anomalies were ventricular septal defects, hypoplastic left heart and tetralogy of Fallot.

Hoffman JIE (2002)[21] stated that the incidence of congenital heart disease in different studies varies from about 4/1000 to 50/1,000 live births. Ventricular septal defects were the most common type of congenital heart diseases. Study conducted by Barbosa JM (2002)[10]estimated the incidence of cardio vascular anomalies as 8 per every 1000 cases.

In the study of Hui L (2005)[24] incidence of cardiovascular defects was 4.68% and anomalies defected were Tetralogy of Fallot, common atrial truncus, double outlet right ventricle, transposition of great vessels. Similar study was carried out by Archer man RJ (2007)[1]. Incidence of cardiovascular system anomalies in their study was 2 per 1000 cases.

Sipek A (2010)[53] conducted the study on 1,472,610 live births. Congenital cardiac anomalies were the most common birth defects (40%). As a whole 29,133 congenital cardiac defects were diagnosed (198.88 per 10,000 live births or 19.9 per 1000 live births).

In the present study the incidence of cardiovascular system anomalies is 3 per 1000 pregnancies. Cardiovascular anomalies are 9.7% of all anomalies detected during the study. These findings are comparable with the study of Narchi H (1997)[43]andArcherman RJ (2007)[1]. In the present study all of the cases are ventricular septal defects. This finding is comparable with Hoffman JIE (2002)[21].

Table 11: Congenital diaphragmatic hernia

Investigators	Incidence
Skargard ED (1999)[54]	1 per 2000
Dillon E (2000)[16]	1 per 4000
Phatak SV (2006)[46]	1 per 2000
Present study	1 Per 1000

Study on congenital diaphragmatic hernias and their antenatal detection were carried out by Skargard ED (1999)[54], Dillon E (2000)[16], Phathak SV (2006)[46]and Gallot D (2007)[18]. During the study conducted by Skargard ED (1999)[54] the incidence of congenital diaphragmatic hernia was estimated to be 1 per 2000 to 5000 births. In the study of Dillon E (2000)[16] the incidence of congenital diaphragmatic hernia was estimated as 1 per 4000 births, which were detected antenatally. Commonest defect in their study was Bochdalek type of hernia. Study of Phathak SV (2006)[46] estimated the incidence of congenital diaphragmatic hernia as 1 per 2000 to 4000 live births.

Gallot D (2007)[18] conducted the study on 501 cases of congenital diaphragmatic hernia which were identified from a total of 1,835,022 live births.

In the present study, the incidence of fetal diaphragmatic hernia of Bochdalek type is 1 per 1000 pregnancies. Congenital diaphragmatic hernia is observed in 3.2% of anomalies.

The incidence of congenital diaphragmatic hernia in present study is on higher side than previous studies, as in the previous studies the incidence was calculated from live births but in the present study the incidence is calculated during second trimester of pregnancy. The incidence in birth is on lower side because most of the congenital diaphragmatic hernia cases abort spontaneously during antenatal period.

Table 12: Anterior Abdominal wall defects

Investigators	Incidence
Marrow RJ (1993)[41]	1 per 2500
Present study	1 Per 1000

Mann L (1984)[40], Meizner I (1986)[42]and Marrow RJ (1993)[41] conducted studies on fetal anterior abdominal wall defects. Mann L (1984)[40] conducted a study for

prenatal assessment of anterior abdominal wall defects and 88 cases of abdominal wall defects with and without other lesions were studied. Those were 10 cases of body stalk anomaly, 16 Gastroschiasis (37.5%) and 62 exomphalos (53%). During the study of Meigzner I (1986)[42] 9 abdominal wall defects were observed: Omphalocele (n=5), Gastroschiasis (n=1), cloacal extrophy (n==1) and Prune belly syndrome (n=2). In the study of Marrow RJ (1993)[41] the incidence of abdominal wall defects is calculated as 1 per 2500 cases. These defects included Gastroschiasis, Omphalocele and Body Stalk anomalies.

In the present study the incidence of fetal anterior abdominal wall defects is 1 per 1000 pregnancies. The anterior abdominal wall defect found in the present study is Gastroschiasis. Anterior abdominal wall defects are calculated as 3.2% of fetal anomalies. Incidence in the present study is on higher side than previous studies as in the present study anomalies are observed in fetuses of second trimester of pregnancy.

Table 13: Gastro intestinal tract anomalies

Investigators	Incidence/1000
Haeusler MC (2002)[19]	0.5
Tan HH (2003)[55]	0.5
Present study	1

Study on gastrointestinal tract obstructions were carried out by Haeusler MC (2002)[19]and Tan HH (2003)[55]. Hausler MC (2002)[19] found 349 gastrointestinal obstructions out of 6,70793 cases. He calculated the incidence of gastrointestinal obstruction as 0.5 per 1000 cases. Cases found in their study were oesophageal obstruction and large intestinal obstructions. Amongst those duodenal obstructions were common. Another study of gastrointestinal obstruction was carried out by Tan HH (2003)[55]. They found out the incidence of gastro-intestinal obstructions as 0.5 per 1000 cases.

In the present study, one case of duodenal obstruction is found. The incidence of gastrointestinal obstruction is calculated as 1 per 1000 pregnancies. Incidence in the present study is higher than previous studies of Haeusler MC (2002)[19]and Tan HH (2003)[55]. In the previous study the incidence was measured in live births while in the present study the incidence is calculated in the pregnancies. Incidence of gastrointestinal defects are on the higher side in the pregnancies than the live births as most of the gastrointestinal anomalies are aborted spon taneously. Gastrointestinal tract obstructions are seen in 3.2% of fetal anomalies.

Simple ovarian cyst

Study on the fetal ovarian cyst was carried out by Kwak DW (2005)[31]. During their study 17 cases of simple ovarian cysts were studied, 10 fetuses had those cysts on right side, 5 fetus had it on left side and 2 had bilaterally. Present study shows 3 cases of simple ovarian cysts. Out of these three cases one is having cyst on right side and remaining two cases are having it on left side. Incidence of simple ovarian cyst in the present study is 3 per 1000 pregnancies. Simple ovarian cysts are observed in 9.7% of fetal anomalies.

Table 14: Fetal urinary system anomalies

Investigators	Percent of anomalies
Isaksen CV (2000)[25]	27
Scott JES (2002)[49]	19.6
Present study	9.7

Fetal urinary anomalies were studied by Isaksen CV (2000)[25], Scott JES (2002)[49], Hosseiniparah SM (2007)[23]. In the study of Isaksen CV (2000)[25] urinary system anomalies were giving an incidence of 27% of other anomalies. During the study of Scott JES (2002)[49]

the incidence of fetal urinary anomalies were 19.6%. Hosseiniparah SM (2007)[23] conducted research with the aim of studying the incidence of urinary malformations. From 774 cases of autopsies 33 cases were urinary malformations which made 4.26% of all cases.

In the present study, urinary system anomalies are 9.7% of all other anomalies and incidence of urinary anomalies is 3 per 1000 pregnancies. When we compare our study with the studies conducted by Isaksen CV (2000)[25]and Scott JES (2002)[49] the incidence of urinary system anomalies were on the lower side as the studies conducted by Isaksen CV (2000)[25]and Scott JES (2002)[49] included fetal, perinatal and infant population but the present study includes fetuses in the second trimester of pregnancy.

Musculoskeletal anomalies

Prenatal sonographic study of fetal musculoskeletal anomalies was carried out by Rvu JK (2003)[47]. They encountered 50 cases of focal musculoskeletal anomalies. Those were including focal limb dysplasia, mesomelic dysplasia, anomalies of hand, anomalies of foot, amniotic band syndrome and anomalies of spine. Similar study on prenatal diagnosis and treatment of congenital anomalies of hand and upper limb was carried out by Bae DS (2009)[8]. During their study the incidence of fetal musculoskeletal anomalies was calculated as 6 of every 1000 pregnancies.

In the present study incidence of musculoskeletal anomalies is 4 per 1000 pregnancies. These anomalies includes Talipus – equinovarus (n=1) and short limb dwarfism (n=3). Musculoskeletal defects are observed in 12.9% of fetal anomalies.

Hydrops fetalis

Sharma S (1999)[50] reported a fetus of 17 weeks with bilateral pleural effusion and large amount of ascitic fluid. Fetal occipital bone revealed a large defect with outpouching of meninges, forming a large meningocele. Fetal subcutaneous oedema of scalp and abdominal wall was also observed. The incidence of non-immune hydrops fetalis is approximately 1 in 1500 to 4000 deliveries MacAfee CA (1970)[37].

In the present study hydrops fetalis is seen in 2 cases giving an incidence of 2 per 1000 pregnancies and gives 6.5% of fetal anomalies. Incidence of hydrops fetalis is on higher side in the present study as it is carried out during second trimester of pregnancy and previous studies were carried out during deliveries.

Syndrome-Pentalogy of Cantrell

Yadav P (2003)[60], Khanna PC (2004)[27], Louis JDS (2006)[36], Hoorn JHL (2008)[22]and Korver AMH (2008)[29] reported cases of pantology of Cantrell. Yadav P (2003)[60] reported a case of Pentalogy of Cantrell associated with encephalocele diagnosed prenatally in the second trimester. Fetus had an anterior thoraco-abdominal wall defect with the contents herniating into the amniotic cavity covered by a thin sac. The sac contained liver and spleen along with few loops of intestine. Umbilical cord was attached to the tip of the omphalocele and demonstrated two vessels (one artery and one vein). The heart appeared normal. An encephalocele was found in the fetal head. The diagnosis of pentalogy of Cantrell associated with encephocele was made.

Khanna PC (2004)[27] reported two cases. First case was having a midline umbilical defect with omphalocele; sternal defect with ectopia Cordis, Scoliosis, Clubfoot, Bilateral cleft lip and midline cleft palate. Another case studied by Khanna PC (2004)[27] had an omphalocele and sternal cleft with ectopia cordis. Louis JDS (2006)[36]

studied a case of Hypoplastic left heart syndrome and an uncharacterized midline abdominal wall defect.

Hoorn JHL (2008)[22] reported two cases with incomplete Pentalogy of Cantrell. First case showed a fetus with bilateral hydrothorax.

ectopia cordis, with ventricular septal defect and a large omphalocele with the heart and liver. Another case showed a fetus with ectopia cordis with ventricular septal defect, large omphalocele with heart, stomach, spleen and liver and scoliosis.

Korver AMH (2008)[29] reported one case of pentalogy of Cantrell. Physical examination showed a purple mass just above the umbilical insertion pulsating with the heartbeat. Echo-cardiography on the first day showed a dextroposition of the heart.

There was a large VSD (Venticular Septal Defect) with left to right shunt. There was a diverticulum of the apex of the left ventricle extending caudally through a diaphragmatic hernia just underneath the skin. Through the omphalocele, the left liver lobe and intestines were visible.

The sternum was anatomically complete but was short. Because of the combination of an abdominal wall defect, a diaphragmatic defect, and a heart defect, the diagnosis of pentalogy of Cantrell was made. The estimated incidence of that malformation was about 1 per 65,000 live births.

In the present study, multiple anomalies are found in one case. Fetus is having single ventral abdominal wall defect with a covering membrane, sternal wall cleft with ectopia cordis, kyphoscoliosis, bilateral pleural effusion, mild generalised subcutaneous edema. Defect is associated with gross polyhydramnios. The condition is diagnosed as Pentalogy of Cantrell.

Table 15: Most common anomalies

Investigators	Most common anomalies
Singh M (1980)[51]	Musculoskeletal anomalies
Himmetoglu O (1996)[20]	Neural tube defects
Todros T (2001)[56]	Musculoskeletal anomalies
Aqrabawi HF (2005)[7]	Neural Tube defects
Singh S (2006)[52]	Neural Tube defects
Khattak ST (2008)[28]	Neural Tube defects
Present study	Neural Tube defects

Prenatal diagnosis of congenital anomalies was studied by various investigators and found out most common congenital anomaly.

Singh M (1980)[51] studied the consecutive infants delivered during 1975-78. Almost one-third (31.1%) of all anomalies originated from musculoskeletal system. Himmetoglu O (1996)[20] found neural tube defects as most common anomalies with the incidence 0.27% in their population. Todros T (2001)[56] found that the prevalence of musculoskeletal anomalies were highest. Similarly Aqrabawi HF (2005)[7] found neural tube defects as the most commonly occurred anomaly in their study with the incidence of 6.5 per 1000 live births.

Singh S (2006)[52] conducted the similar study on fetal anomalies. During their study they found neural tube defects as the most common anomalies which was more than 50%. Incidence of neural tube defects was also studied by Khattak ST (2008)[28]. In their study 13.9 per 1000 deliveries of neural tube defects were observed making them as most common anomalies found during the study.

In our study neural tube defects are most common anomalies. The present study is comparable with Himmetoglu O (1996)[20], Aqrabawi HE (2005)[7], Singh S (2006)[52]and Khattak ST (2008)[28] where neural tube defects were most common anomalies.

Conclusion

Today ultrasonography is an important tool for detection of fetal anomalies. As the risk of fetal malformation is present in all pregnant women, many experts believe that prenatal ultrasound screening should be universal.

The project "The study of Fetal Anomalies In Second Trimester Of Pregnancy By Ultrasonography In Central India Region" was carried out in our medical college and hospital, over the period of 2 years.

The study included 1000 pregnant women who were selected from the antenatal care (ANC) patients in second trimester i.e. between 12 and 24 weeks of gestation attending ANC clinic for ultrasonographic screening at our medical college and hospital. The study subjects included rural, urban and migrated population from most part of Central India.

The subjects selected were from the age group of 18 to 40 years. The subjects were given prior appointment in the morning hours and screened under the guidance of only one experienced sonologist throughout the study.

Ultrasound screening of all the subjects was done in detail. Maternal age, menstrual history, obstetric history and drug history were obtained. During ultrasound scanning number of gestation, presentation of fetus, age of the fetus in weeks and days were screened. Placental position was noticed. Amniotic fluid was accessed. Detail screening for fetal anomaly was done.

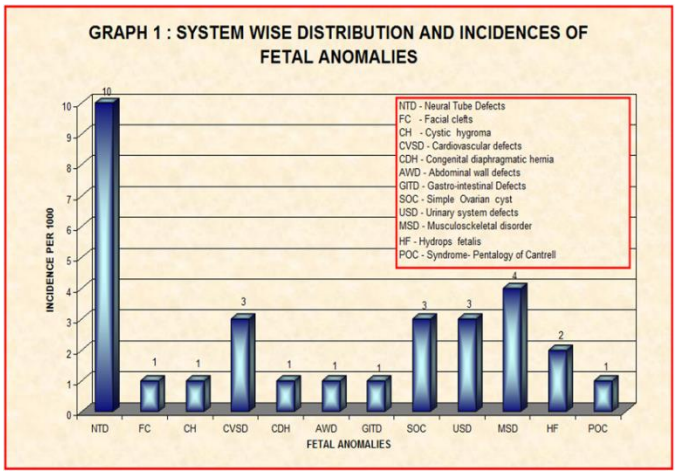
From the above study following conclusions are made.

1. The incidence of fetal anomalies in the Central India Region is calculated as 3.1% of pregnancies in second trimester.

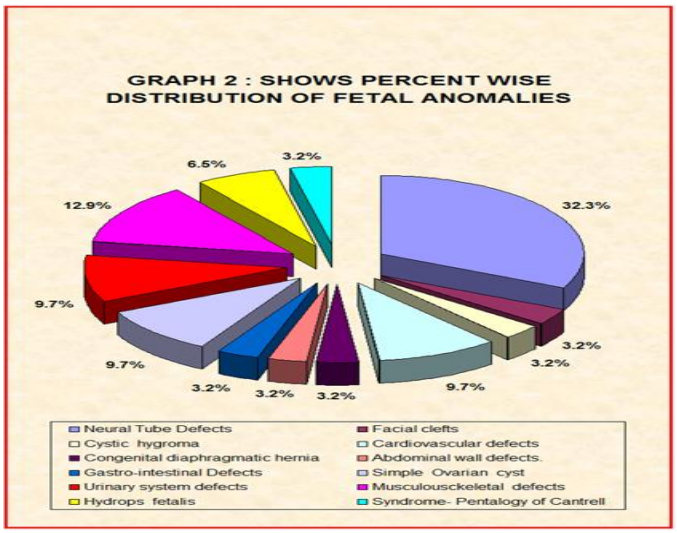
2. During the project following fetal anomalies are studied

- Neural tube defects – Anencephaly (n=3), Holoprosencencephaly (n=2), Aqueductal stenosis (n=2), Hydrocephalus (n=1), Occipital encephalocele (n=1) and Dandy Walker cyst (n=1).
- Facial cleft – Cleft lip on right side.
- Cystic hygroma – Cervical cystic hygroma
- Cardiovascular defects – Ventricular septal defect.
- Congenital diaphragmatic hernia – Bochdalek Hernia on left side.
- Anterior abdominal wall defects – Gastroschiasis.
- Gastrointestinal defects – Duodenal atresia.
- Simple ovarian cyst.
- Urinary defects – Fetal polycystic kidneys with hydronephrosis.
- Musculoskeletal defects – Congenital Talipus Equinovarus and Short limb dwarfism.
- Hydrops fetalis.
- Pentalogy of Cantrell syndrome.
- Most commonly seen fetal anomalies are neural tube defects with the incidence of 10 per 1000 pregnancies which comprises 32.3% of anomalies.
- None of the fetal anomalies are associated with antenatal drug treatment.

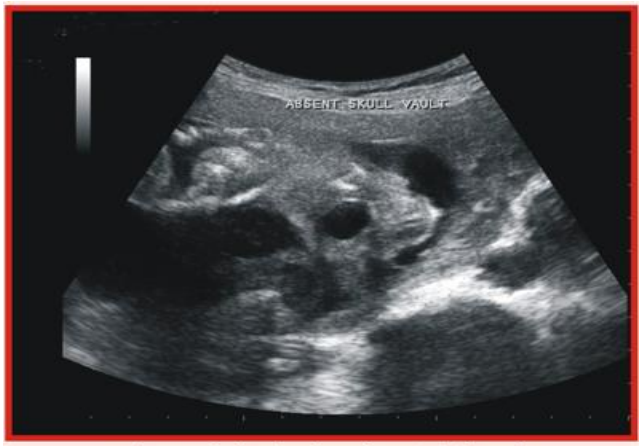
Legend Figure



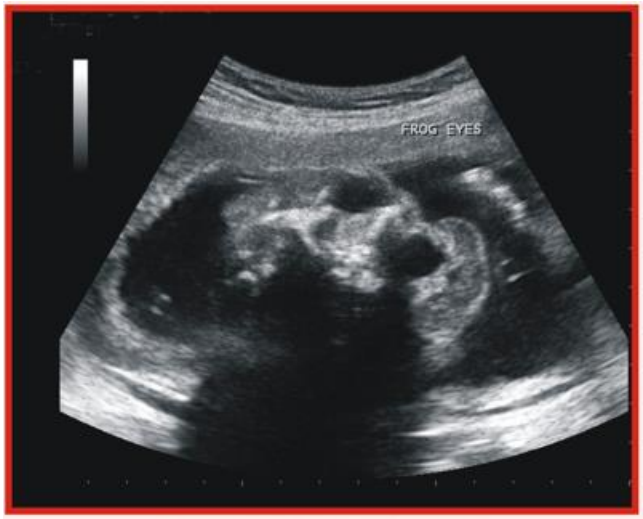
Graph 1: system wise distribution and incidences of fetal anomalies.



Graph 2: shows percent wise distribution of fetal anomalies.

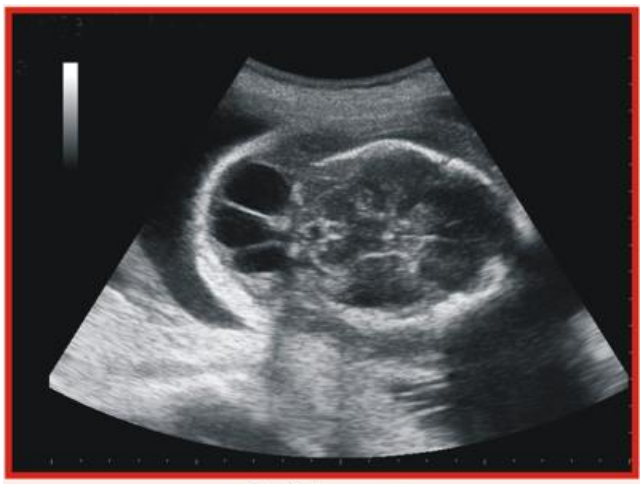


Anencephaly showing absent skull vault

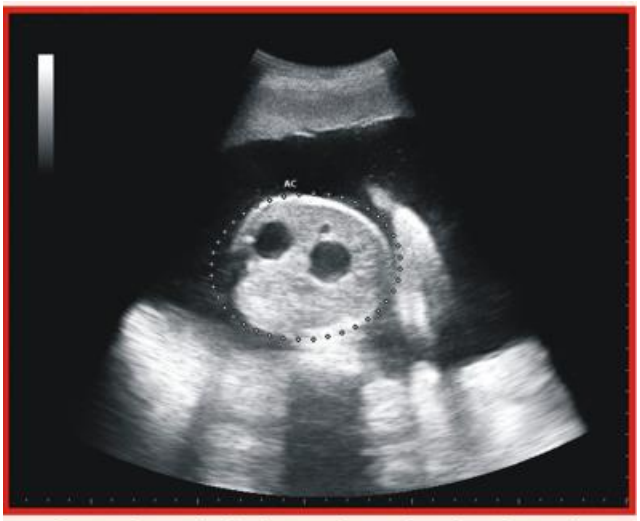


Anencephaly showing frog eyes

Figure 1

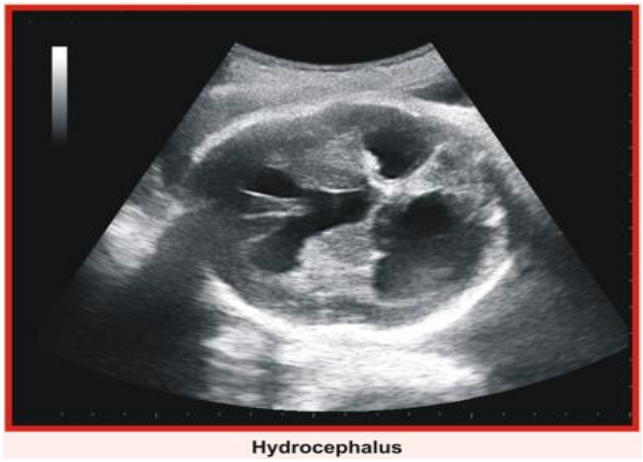


Cystic hygroma

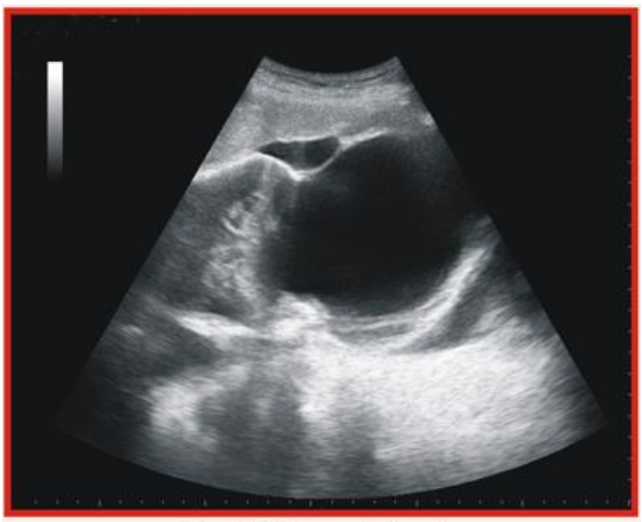


Gastrointestinal obstruction - duodenal atresia

Figure 2

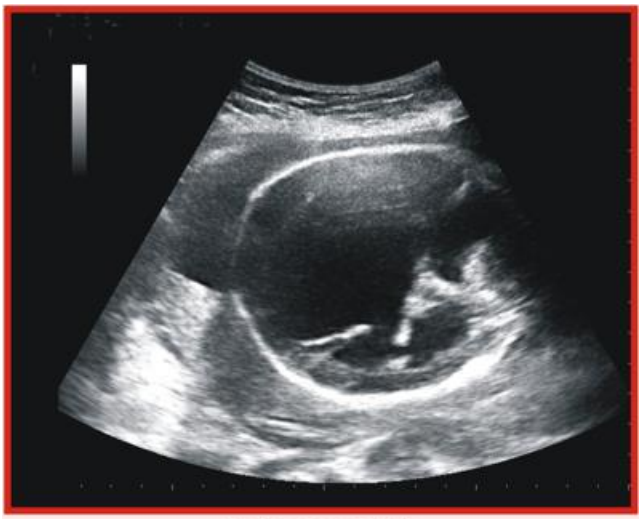


Hydrocephalus

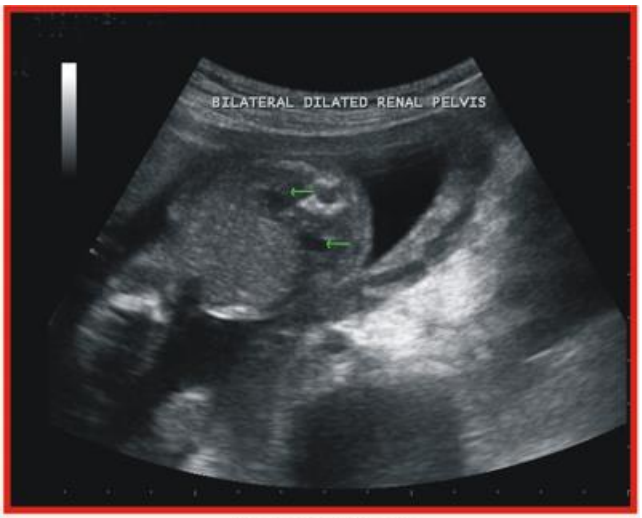


Occipital encephalocele

Figure 3



Holoprosencephaly



Hydronephrosis

Figure 4