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Fetal Anomalies on Antenatal Ultra Sound Scan - A Descriptive Study

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ABSRACT

To evaluate the incidence of congenital fetal malformations by ultrasound in the 18–22 weeks' scan. To describe the fetal anomalies detectedonantenatal Ultrasound studies were done when appropriate. The final outcome of the pregnancy was assessed. Among the 55 antenatal mothers with anomalous fetuses, 13 fetuses were classified as lethal. The single most common anomaly is Anencephaly. The most common organ system involved is Central Nervous System. most of the lethal anomalies are seen in central nervous system. Awareness about fetal karyotyping and fetal autopsy is very less among the patients and a positive step towardspaper highly desirable. Trimester anomaly scan should be offered to all the antenatal mothers between 18-22weeks and should be made an essential part ofthestandard antenatal care.

Keywords: karyotype, fetal autopsy, ultrasound and antenatal care.

1. INTRODUCTION

Congenital anomalies are defined as structural defects, chromosomal abnormalities, inborn errors of metabolism and hereditary diseases diagnosed before, at, or after birth 1. Any deviation from the normal range during morphogenesis constitutes an anomaly2. The incidence of fetal anomalies mentioned in various studies is 0.8 to 5%, approximately 1:150 live births 3. Congenital anomalies account for 8 –15% of perinatal deaths and 13 –16% of neonatal deaths in India 4.

Ultrasound has a great potential in screening for morphological abnormalities throughout all trimesters of the pregnancy, being non-invasive, fast, safe, accurate and reproducible with real time display, causing no discomfort to the patient at any time of gestation 5. A routine mid-trimester ultrasound scan is often performed between 18 and 22 weeks of gestation and many structural abnormalities in the fetus can be reliably diagnosed 6 .This short interval has been chosen because: the development of fetal organs is almost complete; the quantity of amniotic

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fluid is more than the fetal body, allowing a good acousticwindow for penetration of the ultrasound beam; and, if a fetal malformation is detected, it is still possible to plan other diagnostic procedures, such as amniocentesis, or offer the woman the option of terminating the pregnancy in the case of a severe anomaly. Ultrasonography can identify at least 35 – 50% of major fetal malformations with a specificity of 90 – 100% 7.

This study is the effort to detect congenital anomalies with the help of ultrasonography at 18 – 22 weeks of pregnancy. The detection of nature of congenital anomalies can help in deciding the treatment protocol for planning the delivery and also for keeping all the necessary assistance ready for managing the newborn.

2. METHODOLOGY

A prospective study consisting of 2839 antenatal women was conducted in 'SreeBalaji Medical College & Hospital', Chennai, after obtaining ethical clearance from 'Institutional Human Ethical Committee'.

Duration of study: The study was conducted over a period of 19 months from march 2017 to september 2018.Equipment used: In all cases transabdominal ultrasound examination was done using transducer of3.5 MHz frequency which were performed by using an DC -7 unit (Mind ray) Ultrasound machine.

Inclusion criteria: All singleton pregnant women who were referred to the Department of Radiodiagnosis 'SreeBalaji Medical College & Hospital', Chennai, for a second trimester complete antenatal ultrasound examination.

Exclusion criteria: Multiple gestations.

A detailed history of the patient was taken. Risk factors of having fetal abnormality were noted. A detailed systemic and obstetric examination was made. All preliminary investigations were done.Antenatal women between 18 -22 weeks of gestation were offered counseling before the screening. During the counseling, the patients were made aware of the benefits of ultrasound at 18-22 weeks of gestation.

Women were counseled about the interpretation of the results of the screening procedure. After counseling, Written Consent was taken and detailed filling of FORM F under PNDT act was done and were subjected to a mid-second trimester antenatal ultrasound examination using gray scale & color duplex imaging. The scans were carried out by the trained radiologist.Complete information about the gestational age, placental location, fetal biometry & fetal anomalies was collected and tabulated. Previous antenatal scan was also reviewed. This data was correlated with the pregnancy outcome and appropriate statistical analysis was performed. If lethal anomaly

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detected in ultrasound, counseling and option of termination of pregnancy was given. If no lethal anomaly was identified, pregnancy was continued till term and delivered. After delivery, the baby was evaluated for anomalies by the pediatrician and appropriate investigations were done.

3. RESULTS AND DISCUSSION

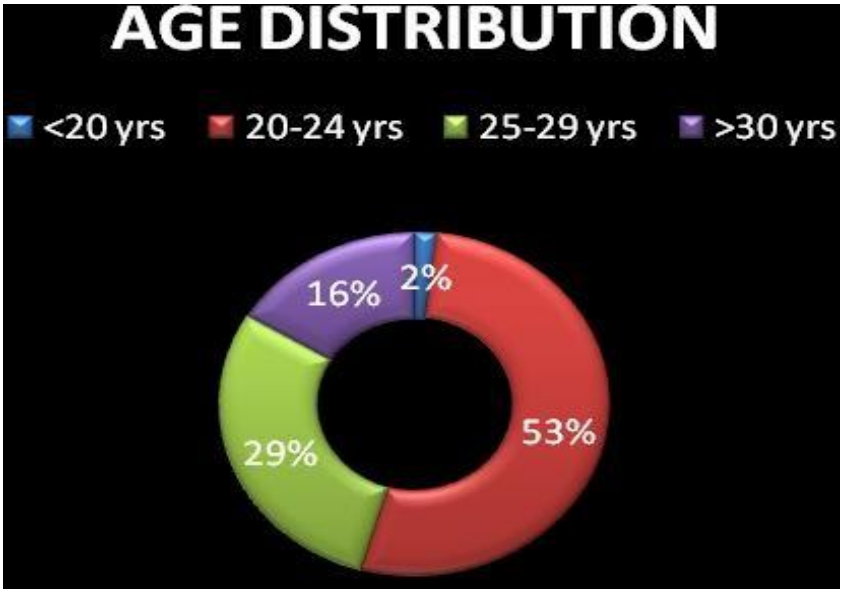
Out of the 2839 cases screened, a total of 55 fetuses was detected to have various anomalies.Prevalence of congenital malformations in the studied population was 1.94 %.

Prevalence (%) = total number of anomalies / total number of scans done x 100

Table1: Maternal Age-wise distributionoffetal anomalies

S. No.	Age Group	No. of Cases	Percentage
1	< 20	1	2%
2	20 - 24	29	53%
3	25 - 29	16	29%
4	> 30	9	16%

FIG 1: Maternal age wise distribution of anomalous fetus.



Majority of the antenatal mothers in the study belong to age group between 20 –24 years. They constituted 53% of the study population. 16 % of the total study population were elderly

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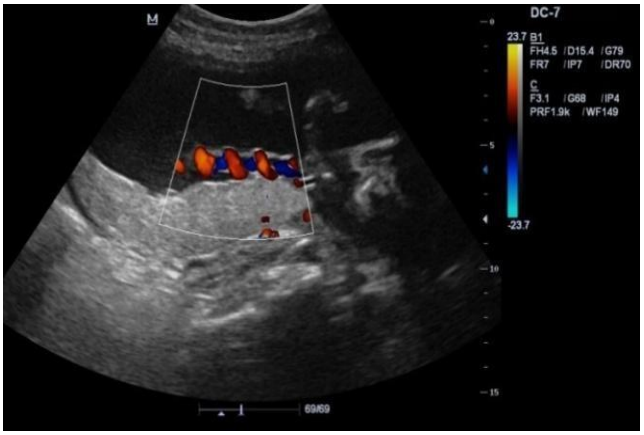
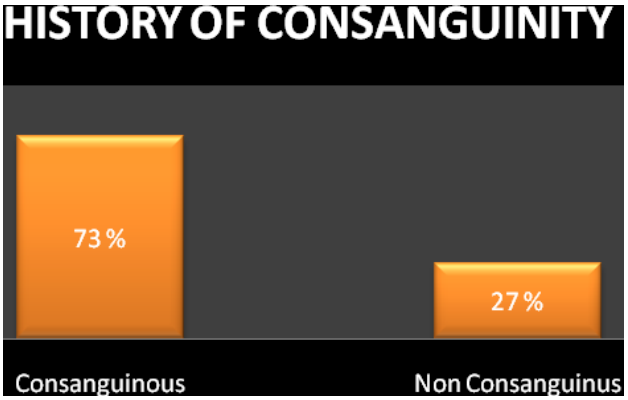
prime.

FIG 2: Single Umbilical ArteryHoloprosencephaly



Percentage of anomalies in primiparaous women (56%) was higher than in multiparaous women (44%).

FIGURE 3: History of Consanguinity amongMotherswithAnomalous Fetuses



Among the 55 fetuses with positive findings for anomalies, 40 mothers (73 %) had history of consanguinity.Majority of the anomalies detected by Ultrasound examination were involving Central Nervous System. In the study population, 22 antenatal mothers had anomalous babies (40%) with CNS involvement. Next most commonly affected is face & neck with 11fetuses (20%). Around 14.5% had genitourinary anomalies. Musculoskeletal anomalies were found in 12.7 % of fetuses. Thoracic anomalies were noted in 7.2% of the fetuses. Anomalies involving multiple systems was detected in 3 fetuses (5.4%).8,9

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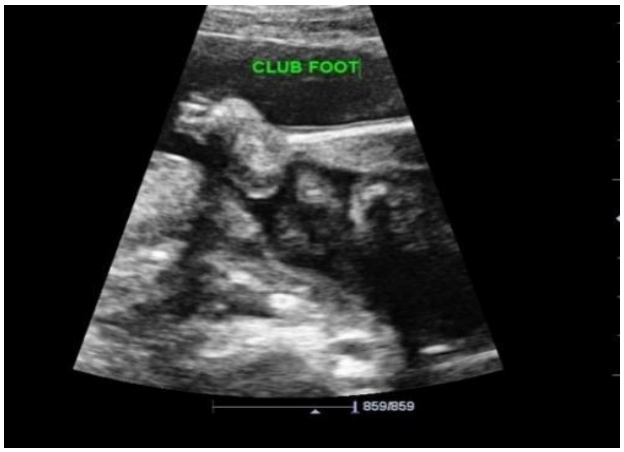
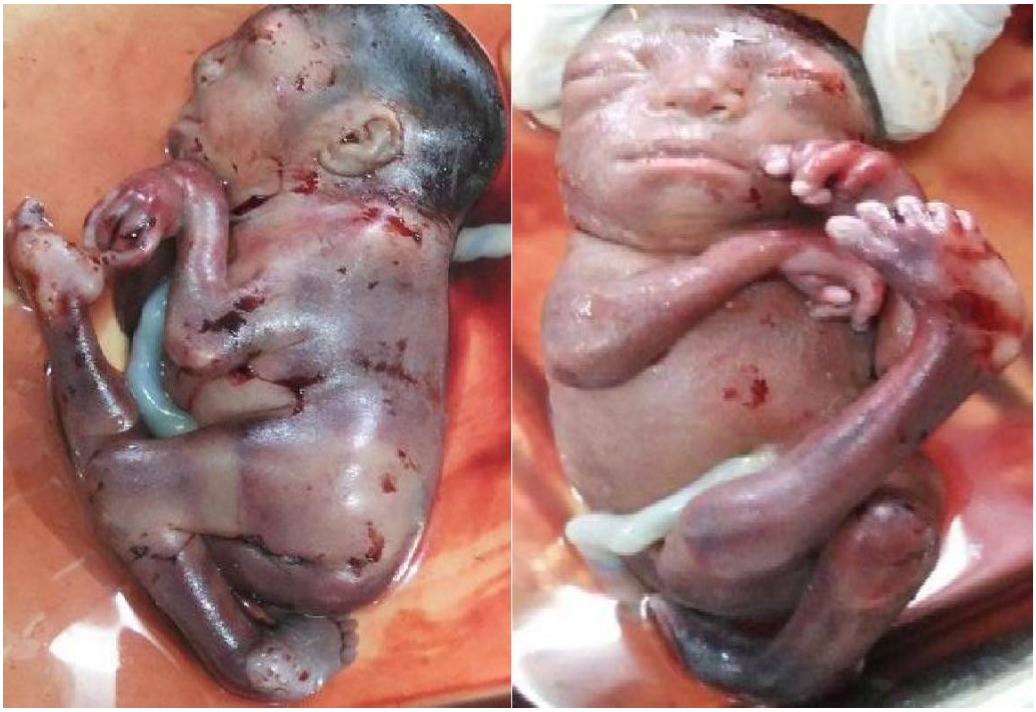
FIGURE 4: SHOWS CLUB FOOT



Analysis of involvement of Organ Systems among anomalous fetuses

FIG 5: Shows Babywasterminatedandultrasoundfindingswere confirmed



A 25-year-old pregnant woman with history of second degree consaguinaneous marriage came for anomaly scan at 20 weeks of gestation. Patient had history of bipolar disorder and was on regular antipsychotics treatment. Patient stopped antipsychotic medication after confirmation of pregnancy. No other history of familial genetic disorder. In sonographic study at 20 weeks of gestation, multiple fetal anomalies were noticed: Lemon shaped skull, Lumbosacral

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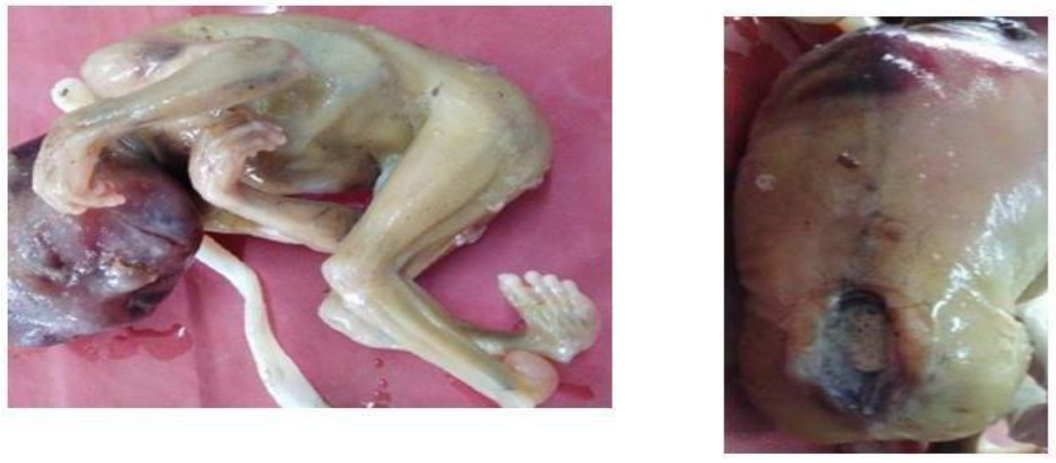
meningomyelocele, Banana sign of cerebellum and obliteration of cisterna magna and Bilateral club foot deformity.Based on these sonographic findings, Arnold Chiari malformation type II was diagnosed. Medical termination of pregnancy was done with delivery of 260 gms dead fetus with deformed scalp, foot with spinal defect. Pregnancy was terminated and sonographic findings were confirmed clinically. Autopsy was not done since parents not giving consent for that.10

Table 2: organ system wise distribution of anomalies

Organ system wise distribution of anomalies			
S.No.	**Organ System involved**	**No. of Fetuses**	**Percentage of anomalies**
1	Central Nervous System	22	40 %
2	Face &Neck	11	20 %
3	Genito-urinary system	8	14.5 %
4	Musculoskeletal System	7	12.7 %
5	Respiratory system	4	7.2 %
6	Cardio vascular system	1	1.8%
7	Multisystem anomalies	2	3.6 %

Figure 6: Banana shaped cerebellum with effaced cisterna magna



Post termination follow up images

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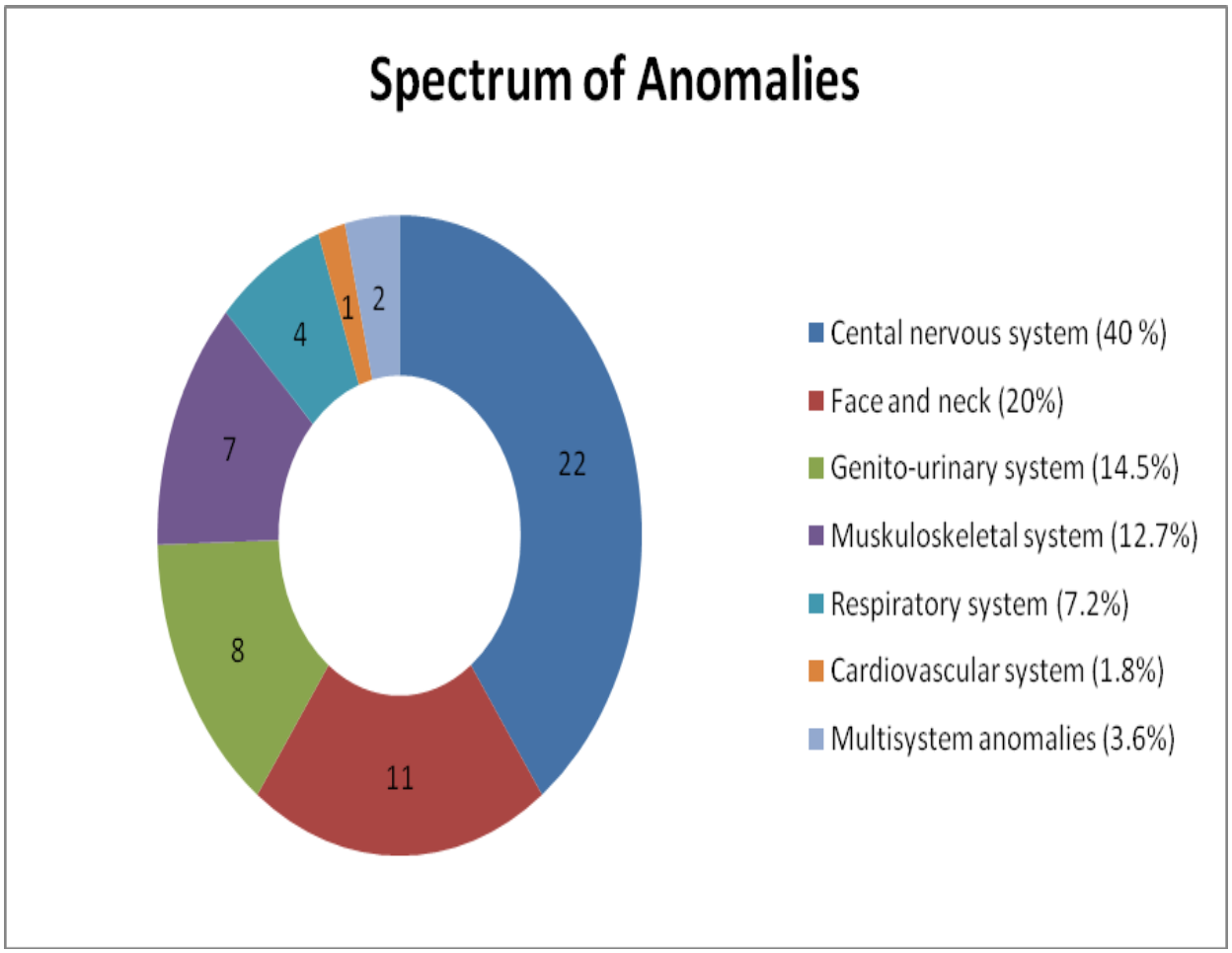
Case 1: A 25-year-old female with history of 20 weeks' gestation came for antenatal scan, with no significant previous past history. No history of consanguineous marriage. Antenatal scan shows a male fetus with evidence of, Severe oligohydramnios, Key hole urinary bladder evidence of posterior urethral valve.

FIGURE 7: ShowingSpectrumofAnomaliesamong Different Systems



Distribution of Central nervous system anomalies in the present study

Among the 22 central nervous system anomalies, we had 6 cases of Anencephaly, 5 cases of ventriculomegaly, 3 cases of Chiari II malformation, 3 cases of Hydrocephalus,2 cases of holoprocencephaly, 1 case of open lip schizencephaly and 1 case of isolated spina bifida.11

Distribution of Face &Neck anomalies in the present study

Among the 11 cases of facial and neck anomalies, we had 6 cases of cleft lip including 2 cases with cleft palate, 1 case with isolated cleft palate, and2 cases of cystic hygroma. The prevalence of cleft lip among the face and neck anomalies is 55%.

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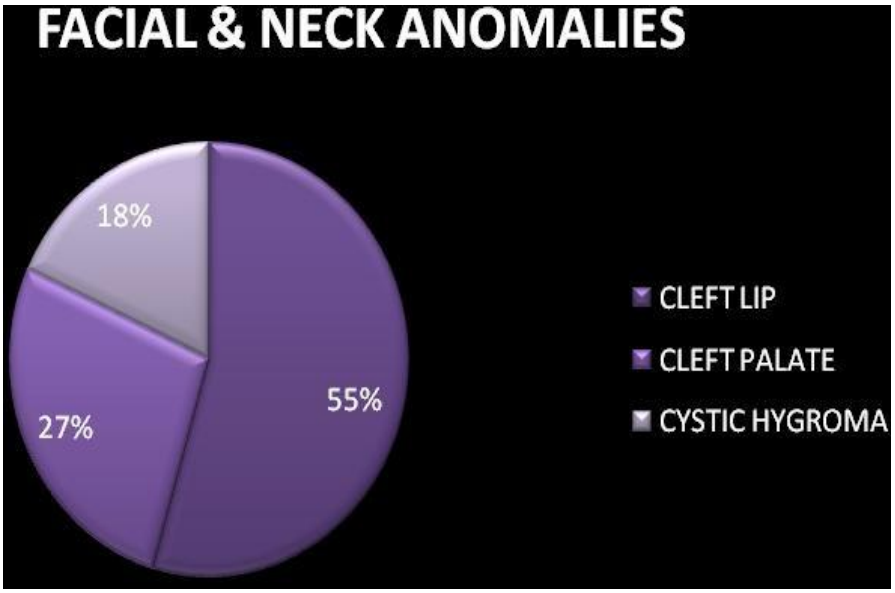
FIGURE 8: Showing Prevalence of Anomalies among Face and Neck



Distribution of genito-urinary system anomalies in the present study

Among the 8 anomalous fetuses with genitor-urinary system abnormalities, 5 cases had multicystic dysplastic kidney, 2 cases had posterior urethral valve and 1 case of renal agenesis.12-15 A 23-year term pregnant woman who got admitted for delivery, came for ultrasound, datingscan and anomaly scan were not done. This present scan shows bilateral cleft lip and cleft palate. No other associated abnormalities identified. The baby was followed post-delivery and the findings were confirmed.

Figure 9: Showing Total Number of Other Anomalies.

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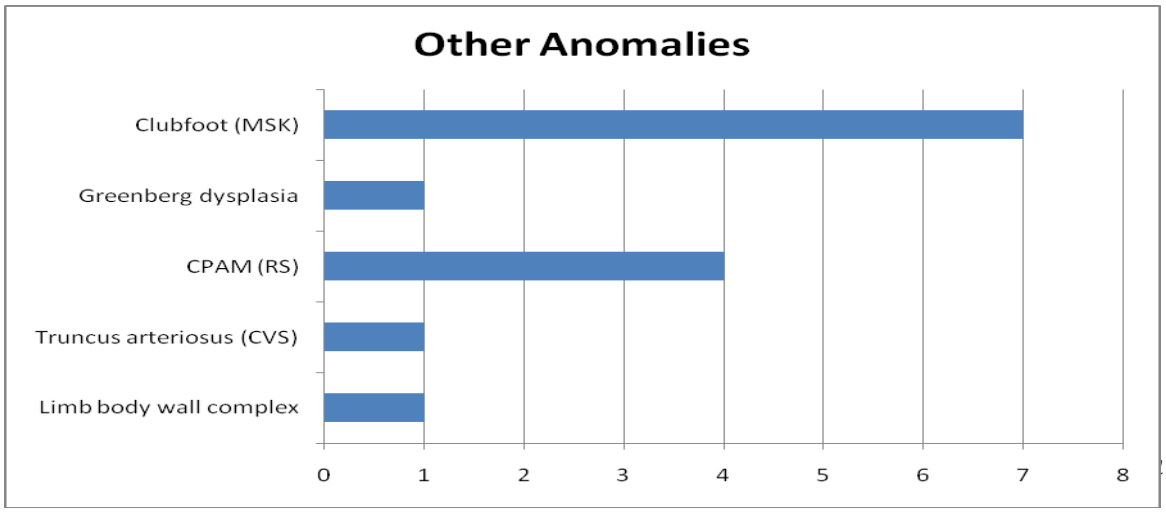
We had 7 cases of club foot among musculoskeletal system anomalies, 1 case of truncus arteriosus among cardiovascular system anomalies, 4 cases of congenital pulmonary adenomatoid malformation, 1 case of Greenberg dysplasia and 1 case of limb body wall complex.



The interpretation and comparison of the results with previous studies are difficult because of varyingcriteria. The present study was limited to determining the incidence and systemic distribution of major anomalies in the specified population. As the present analysis was based on the subjective impression of the author, a true comparison is difficult. Most of the pregnant women referred to the radiology department of our hospital for anomaly scans were in the age group of 20 -24 years and it was noted in our study that the percentage of anomalies was also comparatively more in this age group.14-17

In our study, we observed that more anomalies (73%) were detected in women with consanguineous marriages whereas 27 % of detected fetal anomalies had no history of consanguinity. Prospective study.18 (2017) that the percentage of anomalies in women with consanguineous marriages was higher than women with no history of consanguinity, which corresponded with the results in our study.Out of the 2839 cases screened, a total of 55 fetuses was detected to have various anomalies. Prevalence of congenital malformations in the studiedpopulation was 1.94 %. It is comparable to the prevalence percentage observed in other standard national and international studies. Different authors have reported an incidence ranging from 1.14 to 2.7% in larger series.Central nervous system anomalies.19Dhapate et al (0.19%). S.Singh et al (2006) reported a higher incidence of 0.35%. Anencephaly contributed 27.3% among CNS anomalies in our study. This observation is well within the range observed with larger studies, for instance, studies conducted in India by Dhapate et al showed the incidence to be 48.57%, while Balakumar reported 32.14%.20-23 The diagnosis of Ventriculomegaly remained controversial till Cardoza et al 118(1988) reported that the normal atrial diameter remained

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relatively constant throughout the gestation. Ventriculomegaly is considered mild, if the atrial diameter is 10 to 15mm, moderate if it is 15 to 20mm, and severe if it is greater than 20mm. The main causes of fetal ventriculomegaly are aqueductalstenosis, Chiari II malformation, Dandy-Walker complex, and agenesis of the corpus callosum. In our Study, Ventriculomegaly has accounted for 22.72% of CNS anomalies, as against that observed in the study conducted by Muhammed Nafees et al (36.16%).

In our study, 11 cases of facial clefts were seen. 4 of them had isolated cleft lip and 2 are dianosed with cleft lip and cleft palate and one cae with isolated cleft palate. Maarse et al 119 (2010) reported detection rate of 100% for cleft lip, 86-90% for cleft lip with palate and 0 -89% for cleft palate by 3D ultrasound. They concluded that, the two dimensional ultrasound screening for cleft lip and palate is associated with false-positive results. Three dimensional ultrasound can achieve a reliable diagnosis, but not of cleft palate only.24,25

In our study we had 5 cases with cardiac and respiratory anomalies. 4 cases of congenital pulmonary adenomatoid malformation and 1 case of truncus arteriosus. The prevalence is 0.2 % which is less when compared with the previous work done by Mohammed Nafees et al (2006).

4. CONCLUSION

Ultrasonography is the real workhorse in Obstetric Imaging. The primary goal of routine obstetric sonography at 18-22 weeks' gestation is to ensure structural normalcy of the fetus and to detect fetal anomaly at a time when legal termination of pregnancy is an option. The approach should be 'from ultrasound sign to final diagnosis' and not the other way round. The increasing technical advancements make the radiologists to improve their operator skill. An echo anatomic correlation is desirable. Identifying fetal anomalies at the earliest helps in taking decision by the patient without much emotional involvement. Those who takes decision for termination of pregnancy, it is easier for obstetrician and patient to offer termination. With fetal medicine and multidisciplinary approach, it is possible to provide the unfortunate couple with a reliable estimate of the diagnosis, the cause of the anomaly, the possible treatments if available, the chances of survival and the possibilities of recurrence in subsequent pregnancies.

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Ethical approval: The study was approved by theInstitutional Ethics Committee

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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