

Prevalance of Congenital Fetal Malformations by Ultrasound in the 18 – 22 Weeks Scan

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Abstract

Original Research Article

Congenital anomalies have emerged as an important cause for neonatal morbidity and mortality. The prevalence as well as pattern of anomaly varies from place to place. This is a prospective follow-up study involving 2839 singleton pregnant women who were referred to the Department of Radiodiagnosis of Sree Balaji Medical College & Hospital, Chennai, for a complete second trimester antenatal ultrasound examination. Out of the 2839 cases screened, a total of 55 fetuses were detected to have various anomalies. Prevalence of congenital malformations in the studied population was 1.94 %. Routine antenatal anomaly screening with ultrasound should be done for all patients so that early termination of lethal anomalies can be done.

Key words: Ultrasound; anomalies; prevalence; antenatal; congenital.

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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 anomaly [2]. 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 fluid is more than the fetal body, allowing a good acoustic window 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 the incidence of congenital anomalies with the help of ultrasound 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.

Aims and objectives of the study

- To evaluate the incidence of congenital fetal malformations by ultrasound in the 18 – 22 weeks scan.

MATERIALS AND METHODS

A prospective study consisting of 2839 antenatal women was conducted in 'Sree Balaji 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 of 3.5 MHz frequency which was performed by using a DC -7 unit (Mind ray) Ultrasound machine.

Inclusion criteria

All singleton pregnant women who were referred to the Department of Radiodiagnosis 'Sree Balaji 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 (if any) was collected and tabulated. Previous antenatal scan were also reviewed. This data was correlated with the pregnancy outcome and appropriate statistical analysis was performed.

If lethal anomaly 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.

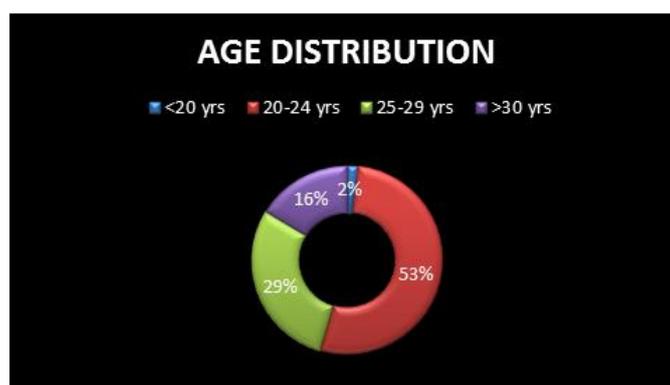
RESULTS

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

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

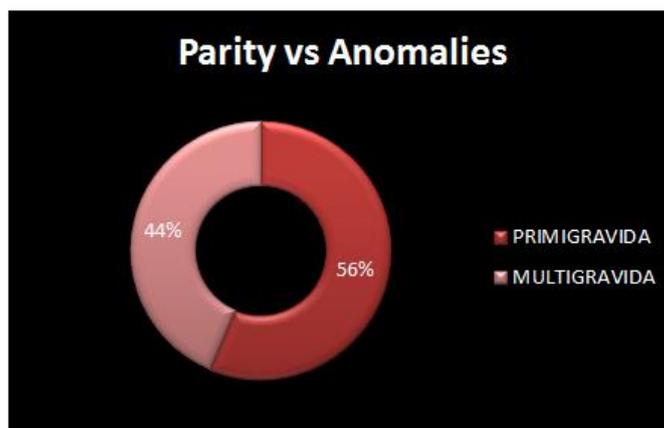
Table-1: Maternal Age-wise distribution of fetal anomalies

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%

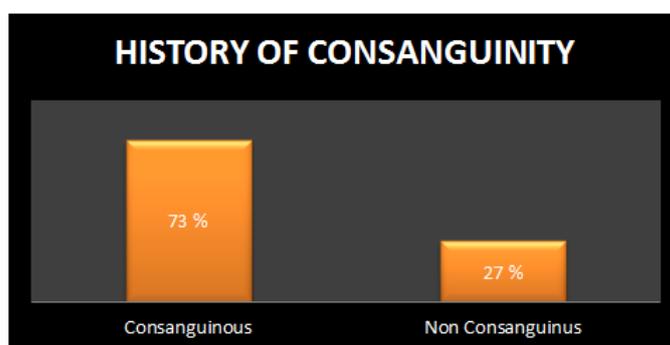
**Graph-1: Maternal age wise distribution of anomalous fetus**

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 was elderly primi.



Graph-2: distribution of antenatal women with anomalous fetus according to parity
 Percentage of anomalies in primiparaous women (56%) was higher than in multiparaous women (44%).



Graph-3: history of consanguinity among mothers with anomolous fetuses
 Among the 55 fetuses with positive findings for anomalies, 40 mothers (73 %) had history of consanguinity.

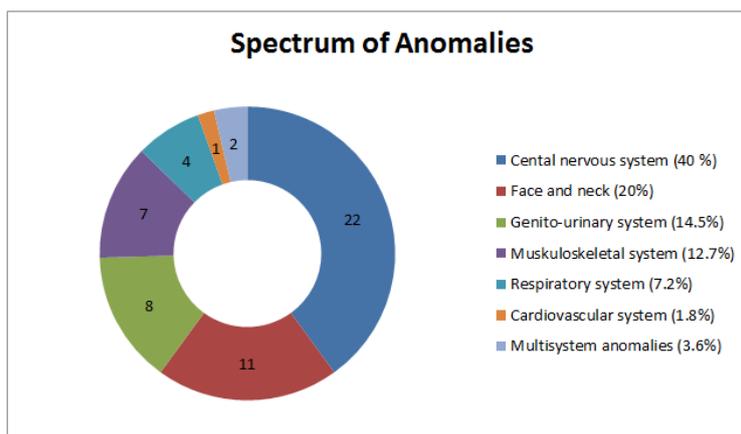
Analysis of involvement of Organ Systems among anomalous fetuses

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 were detected in 3 fetuses (5.4%).

Table-2: 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 %

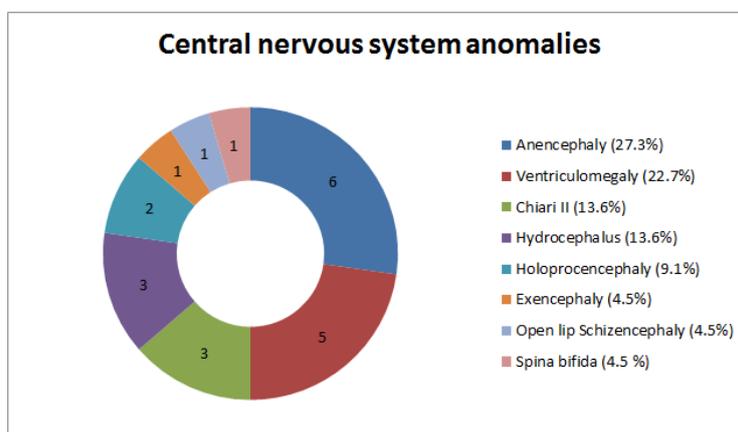


Graph-4: Showing spectrum of anomalies among different systems

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.

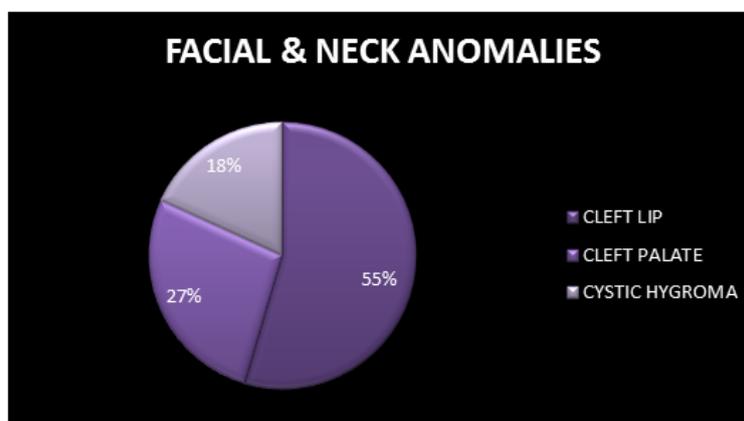


Graph-5: showing prevalence of various central nervous system anomalies

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, and 2 cases of cystic hygroma. The prevalence of cleft lip among the face and neck anomalies is 55%.

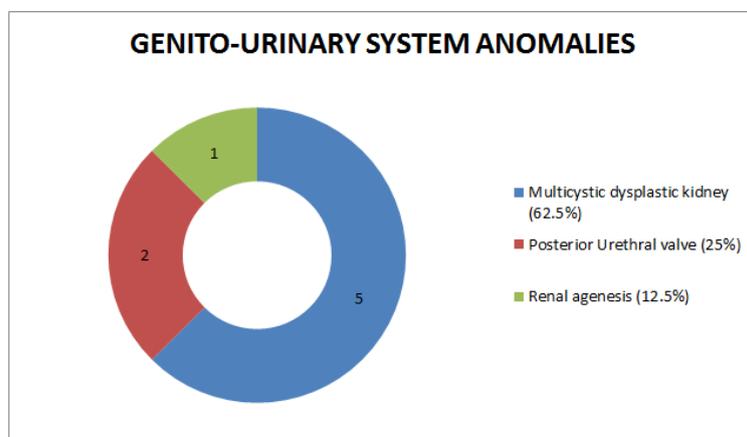


Graph-6: showing prevalence of anomalies among face and neck

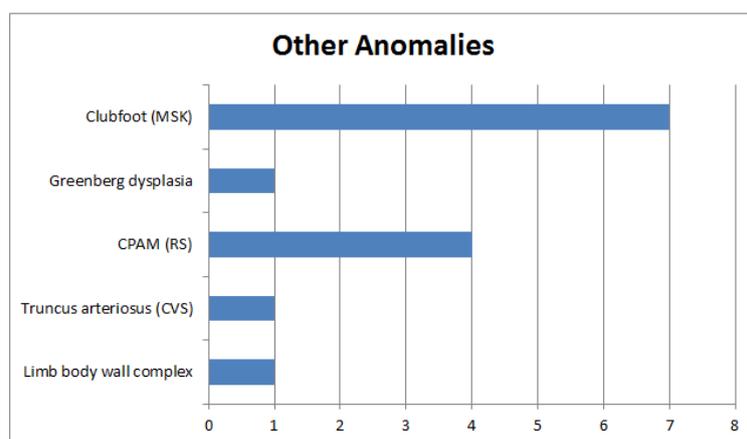
Distribution of genito-urinary system anomalies in the present study

Among the 8 anomalous fetuses with genito-urinary system abnormalities, 5 cases had multicystic

dysplastic kidney, 2 cases had posterior urethral valve and 1 case of renal agenesis.



Graph-7: Prevalence of various anomalies among genito-urinary system



Graph-8: Showing total number of other anomalies

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.

DISCUSSION

The interpretation and comparison of the results with previous studies are difficult because of varying criteria. 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.

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 by Lakshmi Prabha Subhash R *et al.* [8] 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.

In our study, we observed that more anomalies (56 %) were detected in primipara whereas 44 % of detected fetal anomalies were multipara. Prospective studies by Amar Taksande *et al.* [9] and Singh *et al.* [10] found that the percentage of anomalies in primipara women was higher than in multipara, which corresponded with the results in our study.

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

Table-3: Comparison of Incidence of Fetal Malformations with other Studies

Author/Year	Year/Country	No. of Cases	prevalence
Levi S <i>et al.</i> [10]	1991/Belgium	16,370	2.3%
Ewigman BJCJ <i>et al.</i> [11]	1993/USA	7,812	2.4%
Julian Reynier <i>et al.</i> [12]	1994/France	1,64,509	1.26%
Grandjean <i>et al.</i> [13]	1999/Europe	1,70,800	2.7%
Singh S <i>et al.</i> [14]	2006/India	10,890	1.14%
Balakumar K [15]	2007/India	30,030	2.59%
Present Study, 2018	India	2,839	1.94%

Central nervous system anomalies

The central nervous system anomalies was the commonest involved in this study with a prevalence of 0.7 % and forming 40% of the anomalies among the 55 anomaly cases. Weston *et al.* [11] reported similar findings.

Among the central nervous system anomalies the neural tube defects are more. Anencephaly was the most common neural tube defect in this study. Incidence of anencephaly in our study was 0.2%. This incidence is in line with the study conducted by Dhapate *et al.* (0.19%). S.Singh *et al.* 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% [12]. The diagnosis of Ventriculomegaly remained controversial till Cardoza *et al.* [13] reported that the normal atrial diameter remained 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 aqueductal stenosis, 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%).

Face and neck Anomalies

In our study, 11 cases of facial clefts were seen. 4 of them had isolated cleft lip and 2 are diagnosed with cleft lip and cleft palate and one case with isolated cleft palate. Maarse *et al.* [14] 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.

Genitourinary system

Prevalence of Genitourinary anomalies in our study was 0.4 % forming 14.5 % among the 55 anomalous fetuses. It is well within the range of 6.4 to 20.7% observed in various national and international articles. A comparison of the same is tabulated below:

Table-4: Comparison of incidence of genitourinary anomalies with other studies

S. No.	Author	Year	Incidence Percentage
1	Mohammed Nafees <i>et al.</i>	2006	13.4%
2	Balakumar	2007	18.1%
3	S. Singh <i>et al.</i>	2006	6.4%
4	Euro fetus study group	1999	20.7%
5	Present study	2011	14.5 %

Musculoskeletal System

Congenital Talipes Equino Varus is the only isolated musculoskeletal anomaly noted in the present study with 7 cases with an incidence of 0.2%. 3 cases

had history of consanguinity and 2 cases had bilateral club foot. The incidence of CTEV was 0.2% in our study. Swamy *et al.* reported an incidence of 0.06% out of 75,933 pregnancies.

Table-5: Comparison of Incidence of Musculoskeletal Anomalies with other studies

Sl.no.	Author	No. of pregnant women studied	Incidence among pregnancies
1.	Swamy <i>et al.</i>	75,933	0.06%
2.	Our study	2839	0.2%

Cardiac and Respiratory Anomalies

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.*

Table-6: Comparison of incidence of cardiothoracic anomalies among anomalous babies with other studies

Sl.no	Author	No. of anomalous babies	Incidence among anomalous babies
1.	Mohammed Nafees <i>et al.</i>	134	0.7%
2.	Our study	55	0.2%

CONCLUSION

Incidence of antenatally diagnosed congenital anomalies is increasing as compared to the past because of advanced diagnostic facilities and better-trained radiologists. Fetal congenital anomalies are a major cause of perinatal and infant mortality, so routine antenatal ultrasound examination should be performed for all pregnant women to diagnose these anomalies. Regular antenatal visits and prenatal diagnoses are recommended for prevention, early intervention and even planned termination, when needed. Before starting the examination, a healthcare practitioner should counsel the woman / couple regarding the potential benefits and limitations of a fetal ultrasound scan. Although many malformations can be identified, it is acknowledged that some may be missed, even with good ultrasonography equipment in the best of hands, or that they may develop later in pregnancy.

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