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FOETAL ANOMALY SCAN: A STUDY AT 18 TO 24 WEEKS OF PREGNANCY

Anatomy

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ABSTRACT

Background: Foetal anomaly scan is usually carried out between 18 to 24 weeks of pregnancy. This is an ultrasound scan and looks for structural anomalies in the foetus so that parents can be informed about the consequences arising from continuity of pregnancy. Parents have the choice to discontinue or improve the safety of birth.

Aims and objectives: To study the common structural abnormalities detected by routine ultrasound examination at 18 to 24 weeks of gestation.

Methods: The retrospective observational study was conducted at Silchar Medical College and Hospital on 30 cases of congenital malformation from June 2016 till March 2018. Foetal anomaly scan was done on 3000 women in second trimester of pregnancy.

Results: The results of anomaly scan showed that out of 3000 pregnant women between 18 to 24 weeks, 30 pregnant women (1%) had foetal structural anomalies. Out of them, 13 were primigravida and 17 were multigravida. 99% women showed no foetal anomaly.

Conclusion: Prenatal diagnosis of congenital anomalies provides information for decisions during pregnancy and appropriate treatment perinatally and it is assumed to improve perinatal and long term outcome.

KEYWORDS

Foetal anomaly scan, gravida, prenatal diagnosis.

INTRODUCTION:

Congenital anomalies are one of the main causes of morbidity and mortality among the infants. Foetal structural (organs) malformations are seen in 3 to 5 % of all pregnancies[1]. The psychological trauma and cost associated with foetal anomalies has led to use of ultrasound for the prenatal diagnosis as an essential part of antenatal care. Sonographic examination is an accurate, non-invasive, non-ionizing and relatively inexpensive diagnostic tool for determining prenatal anomalies[2]. Early detection of malformation is tremendously improved with the improvement in ultrasound technology offering better resolution. Most congenital malformations occur during third to eight weeks of gestation. The foetus is also susceptible to malformations in stages before and after this so that no period of gestation is completely free of risk. Several malformations occur later than the first trimester of pregnancy. Some transient findings like midgut herniation, small septal defects etc. may get resolved later during intrauterine foetal life[3]. Foetal anomalies can be detected by sonography at 11-14 weeks, but for detailed foetal anatomic survey 18-22 weeks sonography is necessary (the best time would be between weeks 19 and 20). Ultrasound accuracy remains dependent on gestational age and also on the skill of the sonologist. During the last decades, an increasing number of congenital anomalies have been diagnosed prenatally by ultrasonography. Congenital anomalies are higher among the referral centre as compared with the general population. Even today, the benefits of this technology are not in the reach of all. Many patients in India, had their first ultrasonography for anomaly detection after 20 weeks. It reflects the unawareness of knowledge in patients and also in basic health care facility. Some are not yet undergoing anomalies scan.

A wide range of anomalies of central nervous system, cardiovascular system, gastrointestinal system, urogenital system and skeletal system can be diagnosed by the scan. So, the study was carried out in attempt to find out the common structural abnormalities during the second trimester of pregnancy.

AIMS AND OBJECTIVES:

The aim of the present study was to find out the common structural abnormalities detected by routine ultrasound examination at 18 to 24 weeks of gestation. Early detection of malformations was our primary objectives, so that the parents can be informed about the consequences arising from continuity of pregnancy. Parents have the choice to discontinue or improve the safety of birth.

MATERIALS AND METHODS:

The retrospective observational study was conducted at Silchar Medical College and Hospital on 30 cases of congenital malformation out of 3000 pregnancy after approval by Institutional ethical committee, from June 2016 till March 2018. Pregnant women who attended OPD for regular antenatal check-up, underwent routine ultrasound during the second trimester of pregnancy and those having past history of severe foetal anomalies and chronic miscarriages were considered to evaluate for anomaly scan. They were all referred by obstetricians/ gynaecologists for routine work up of pregnancy. All cases complicated by congenital anomalies within 18-24 weeks were included in the study population. Pregnant women before 18 weeks and after 24 weeks were excluded from the present study. Data were collected from patient's files and reviewed for foetal anomalies. The ultrasonographic scans were evaluated with the help of radiologists.

Malformations were classified according to type of malformation, gestational age and organ system involved. Congenital anomalies were system wise divided into five groups according to organ system involved: 1) Central nervous system, 2) Cardiovascular system, 3) Gastrointestinal tract/ abdominal wall, 4) Musculoskeletal and 5) Facial (table 3). Data were analysed statistically in percentage and presented in tabular and graphical form.

RESULTS:

The present study revealed 30 cases of foetal structural malformations during 18 to 24 weeks of gestation amongst 3000 pregnant women. The mothers had an age range of 20 – 42 years. The majority of the anomalies (56.7%) were present in the age group of 30 – 39 years. The frequency of age group wise distribution of cases were shown in table 1. Congenital malformations were observed in terms of the maternal gravid status, 43.3% in primigravida (13 cases) and (56.7%) in multigravida (17 cases) (table2).

Table 1: Frequency distribution of cases to maternal age. (n=30)

Maternal age in years	Number of anomalies	%
< or equal to 20	0	0
21-29	12	40

30-39	17	56.7
>40	1	3.3
Total	30	100

Table 1: showing congenital anomalies are more common in elderly age group (30-39 years).

Table 2: Maternal Parity and frequency of cases. (n=30)		
Parity	Frequency	%
0	13	43.3
1*	6	20
2-4*	11	36.7
Total	30	100

Table 2: shows that congenital anomalies are more common in multigravida (*).

Table 3: Frequency distribution of organ system malformations (n=30)				
Organ system	Anomalies detected	Number of cases	Percentage %	Total percentage
CNS	Anencephaly	7	23.3%	36.7%
	Spina bifida	2	6.7%	
	Holoprosencephaly	2	6.7%	
GIT and abdominal wall	Omphalocele	7	23.3%	26.7%
	Limb body wall complex	1	3.3%	
Skeletal wall	CTEV	5	16.7%	
CVS	Ectopia cordis	3	10%	
Facial	Cleft lip and palate	3	10%	
Total	30	100		

The spectrum of abnormalities is shown in Table 3.

Anencephaly, Spina bifida and Holoprosencephaly were major neural tube defects observed in the present study. Total percentage of central nervous system malformation was 36.7% with an incidence of 3.67/1000 pregnancy (table 3). Seven cases of anencephaly (23.3%) were recorded. The important ultrasonographic features in anencephaly were an absent cranium with an amorphous brain mass to no recognizable brain tissue (Figure 1). Mothers were mostly in the age group of 20 – 29 years and primigravida. In the present study, 6.7% spina bifida (myelomeningocele) were detected in two cases of primigravida and sonographic findings showed failure of fusion of the caudal neural tube. Two cases of Holoprosencephaly (6.7%) were reported, where sonographic findings were non-separation of cerebral hemisphere with a single ventricle (figure 2).

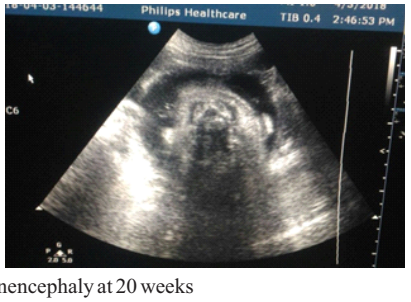

Fig: 1 – Anencephaly at 20 weeks

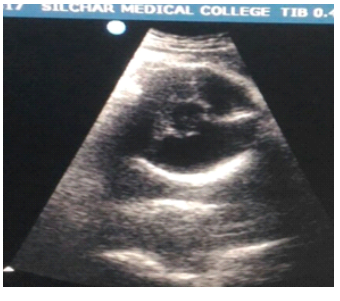

Fig: 2 Holoprosencephaly at 19 weeks

In our study, abdominal wall anomalies were omphalocele (23.3%) and limb body wall complex (LBWC) 3.3% (table 3). Seven cases of Omphalocele (23.3%) were seen in this study, where the maternal age were mostly above 30 years. Prenatal ultrasonographic findings of omphalocele were presence of midline anterior abdominal wall defect, herniated sac associated with visceral contents and umbilical cord insertion at the apex of the sac (figure 3). A 24 years primigravida having a twin pregnancy with a fetus affected with LBWC was reported in our study. Ultrasonography of the affected foetus showed placental attachment to the abdominal wall, wide infraumbilical abdominoschisis, absent umbilical cord, severe scoliosis, absent left leg and malformed right leg (figure 4).

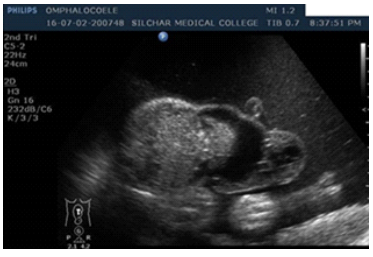

Fig: 3 – Omphalocele at 18 weeks

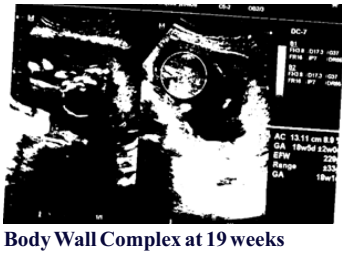

Fig: 4 Limb Body Wall Complex at 19 weeks

Five cases of Congenital Talipes Equino Varus (CTEV) or club foot (16.7%) were observed in this study. Their main ultrasonographic features were presence of one or both feet excessively plantar flexed, with forefoot swung medially and the sole facing inward. Three cases of Cleft lip and palate (10%) were noted and prenatal ultrasonography showed failure of continuity of upper lip and posterior/ anterior portion of the palate using the midsagittal, coronal and axial views of the foetal head and face. Three cases of Ectopia cordis (10%) were observed in our study and were characterized by partial displacement of the heart outside the thoracic cavity.

The most frequent abnormalities detected by ultrasound scan in our study were of central nervous system (36.7%) followed by GIT and abdominal wall (26.7%) as shown in Table 3. In our study, very few (1%) foetuses with malformations were identified in second trimester. Since this was not a prospective study, we did not have the total number of ultrasonography studies performed during the specified period.

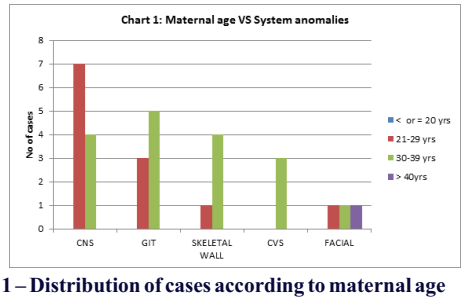

Chart: 1 – Distribution of cases according to maternal age

DISCUSSION:

In our study, very few (1 %) foetuses with malformations were observed in second trimester. This was lower than that reported in the literature for general population 3 to 5 % of all pregnancies[1]. Incidence of congenital anomalies according to Guardiola A et al[4] was 3.67%, Carrera JM5 3.03%, Becker and Wegner[6] 2.8% and Vinodh SL[7] 2.48%. In our study, there may be missing out of an anomaly in many women. It may be due to scarcity of good resolution machines or lack of expertise. Factors that may cause inadequate ultrasound images are poor fetal position, obesity and oligohydromnios.

Maternal age and multigravidas are associated with an increase prevalence of congenital anomalies[8]. A high frequency of congenital malformations in the age group of 30-39 years (56.7%) was seen in our

study. Elderly females need to be examined more carefully since the risk of giving birth to a foetus with congenital anomalies is greater. In our study, multigravida showed increase percentage of congenital anomalies. This was in contrast to a study that demonstrated more congenital anomalies in primipara mothers[9].

In our study, CNS malformations were the most common (36.7%) which was similar with another study3. CNS anomalies are considered the most common in live born and still born foetuses in Egypt and in other countries[10]. Majority of the foetal CNS anomalies are identified by the second trimester ultrasound at 20-24 weeks of gestation, which makes them the most common. CNS anomalies reported in our study were anencephaly, spina bifida (meningomyelocoele) and holoprosencephaly (HPE). Anencephaly is immediately detected when attempting to perform biometry of foetal head. Anencephaly is one of the most common types of neural tube defect, affecting about 1 in 1000 pregnancies. Incidence of anencephaly is 1.1:1000[11](table 4). However, most of these pregnancies end in miscarriage, so the prevalence of this condition in new-borns is much lower (1 in 10,000). In the present study, frequency of anencephaly during second trimester showed 2.3 per 1000 pregnancies. In a study by ZHU Chen, the rate of spina bifida was 1per 10,000 fetuses[12]. HPE occurs in 1/15,000 live births but is present in 1/250 pregnancies that end in early miscarriage[13]. In the present study, frequency of each of spina bifida and holoprosencephaly during second trimester showed 0.7 per 1000 pregnancies. Few authors show an U shaped relationship with maternal age and incidence of CNS congenital anomalies in their neonates, with higher incidence in ages below 20 years or above 35 years[4]. In our study, more cases were seen between 20 – 29 years.

Next common anomalies detected were in relation to anterior abdominal wall. Omphalocele is the most common abdominal wall defect, with a frequency of 1/5000 during pregnancy, decreasing to 0.8/10000 for live births[14]. The incidence of omphalocele is 1/110 foetuses by prenatal ultrasound at a gestational age of 18 weeks, however, this decreases to 1/4,000 live births due to foetal demise[15]. In the present study, frequency of omphalocele during second trimester showed 2.3 per 1000 pregnancies. The diagnosis of omphalocele should not be made before 12 weeks gestation as there is physiological herniation of the midgut during 8–10 weeks and the intestine return to its normal position in the abdominal cavity by 12 weeks. Omphalocele is associated with advancing maternal age, with most of these mothers being over 30 years old[15]. Similar findings were also observed in the present study. Limb body wall complex was first described by Van Allen et al in 1987[16]. It is an abdominal wall defect in addition to a spectrum of limb and visceral anomalies. Present study showed frequency of 0.3 per 1000 pregnancies during the second trimester. It is a rare anomaly with a reported incidence between 1 per 14,000 to 1 per 31,000 pregnancies[17]. The incidence at birth is about 0.32 per 100,000 births because the majority of affected foetus undergo intrauterine deaths[18].

Next common anomaly seen in our study was Congenital Talepes Equino Varus (CTEV). Simultaneous visualisation of leg and foot bones in the same plane is done for accurate diagnosis. Present study showed frequency of 1.7 per 1000 pregnancies during the second trimester. Incidence ranges from about 0.1% in the new born population to 0.4% when diagnosed antenatally by ultrasound[19]. Another study showed the incidence of foetal talipes following routine ultrasound examination was 0.10%[20]. Incidence of CTEV diagnosed before birth varies significantly in published studies, with a range going from 0.43%[20] to 59.8%[21]. Few cases are diagnosed on a late stage of gestation ranging from 22nd to 24th week with initially normal ultrasounds. Such cases almost always correspond to CTEV of a postural origin[21].

Ectopia cordis is a rare congenital anomaly and is detected at 18-23 weeks of gestation. Antenatal foetal anomaly scan at 18 to 24 weeks can detect 50% of the major congenital heart diseases. The prevalence of ectopia cordis has been calculated to be 0.7 to 0.8 per 10,000 deliveries[22]. Present study showed frequency of 1 per 1000 pregnancies during the second trimester. Cleft lip with or without cleft palate is best identified between 20-22 weeks[23]. A study reported an incidence of orofacial clefts is about 1 in 700-1000 deliveries [24]. Present study also showed frequency of 1 per 1000 pregnancies during the second trimester.

The ultrasound failed to scan any renal defects in our study. A thorough knowledge of the ultrasonographic features of normal foetal development is necessary to avoid potential diagnostic pitfalls. Discrepancy between prenatal and postnatal series can be partly explained by the unexpectedly high tendency towards spontaneous intra-uterine demise and early postnatal death of foetuses with severe abnormalities.

Table 4: Comparative data showing incidence of anomalies by various researchers.

Author	Year	Anomalies	Frequency **during pregnancy**	Present Study- frequency per 1000 during 2nd trimester
Chatzipapas IK	1999	Anencephaly	1.1/1000	2.3
ZHU Chen	2016	Spina bifida	1.6/10,000	0.7
Sadler T W	2015	Holoprosencephaly	1/250	0.7
Barisic I	2001	Omphalocele	1/5000	2.3
Paola Linda	2014	Limb body wall complex	1/14,000	0.3
Amato J J	2000	Ectopia cordis	0.7 to 0.8/100000	1
Keret D	2002	Club foot	0.4%	1.7
Melnick M	1990	Cleft lip and palate	1/ 700 to 1000	1

CONCLUSION:

In our study, only 1% of foetuses with malformations are identified in second trimester. CNS malformations were the most prevalent anomaly detected. The present study reveals a high frequency of congenital malformations in the elderly age group and especially among multigravida women. In India, medical termination of pregnancy is done up to 20 weeks of gestation. So, anomaly scan between 18-20 weeks of gestation is essential. A trend toward more pregnancy terminations and fewer new born with anomalies was apparent over the years. Evaluation before conception and counselling even before the pregnancy begins is recommend as preventive measures. Foetal karyotyping should be offered in cases with CNS anomalies to diagnose the genetic cause for further genetic counselling and management. 3D and 4D USG and MRI may help to confirm suspected anomalies detected on routine anomaly scans. Further studies are needed to include more current data to investigate the perinatal mortality or pregnancies with congenital anomalies.

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