EARLY ULTRASOUND DIAGNOSIS OF CONGENITAL ANOMALIES

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SUMMARY

This study proves that majority of the fetal anomalies could be detected at endovaginal sonography performed in and around the 14th week of gestation. Hence we argue that a routine sonographic study by late first trimester and/or early midtrimester should be integral part of prenatal care medicine for detection of fetal anomalies.

By modern standards of Obstetric practice, particularly aiming at 'Preventive Obstetrics', properly spaced three ultrasound studies, in the first, mid and third trimester, should be in order the achieve an excellent pregnancy outcome and to allow the pregnant mother reach term and deliver her baby with confidence.

Detection of anatomic congenital anomalies has been the current theme in prenatal care (Romero et al., 19991). Prenatal diagnosis of congenital anomalies offer several advantages. Many fetal anomalies are not compatible with extrauterine survival, and hence an prenatal diagnosis will allow an earlier and safer pregnancy termination before viability. Other infants may benefit by the diagnosis of anomalies whose prognosis is affected by early recognition and treatment.

Improved resolution of the modern ultrasound equipments combined with the experience and skill of the operator provides for precise detection of fetal abnormalities early in the course of pregnancy (Sabbagha, 1987). Anencephaly, holoprosencephaly, encephalocele, omphalocele, conjoined twins, ventral wall defects, ectopia cordis, and generalized body edema are several of the anomalies noted in the first trimester (Romero et al, 1991, Sabbagha, 1987). None of these conditions are compatible with neonatal survival, and pregnancy termination in first trimester is simple and offers great relief to the woman. Most of the fetal anomalies are diagnosed before the 20th week of gestation,

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except probably the gastrointestinal defects; and hence, routine ultrasound examination should be advocated if the goal is early prenatal diagnosis of fetal anomalies as part of 'preventive obstetrics'.

In this communication the authors are advancing logical arguments in favour of routine ultrasound studies early in pregnancy, and that too level III or level IV sonography, which should be the goal of primary health care in Obstetrics. The arguments are amply supported and well substantiated by the scientific data collected by the authors over a period of one year on 336 first trimester scans and 694 obstetrics scans.

MATERIAL AND METHODS

Endovaginal sonography employing 5 MHz electronic probe has been used for first trimester and early midtrimester (upto 16 weeks of gestation) sonographic studies. Midtrimester gestations beyond 16 weeks have been studied and vaginal probes have been employed as complemetary to each other for pregnancies studies between 16 and 20 weeks. Electronic sector probe with 3.5 MHz frequency has been regularly employed for third trimester scans.

We advocate routine ultrasound evaluation of all pregnancies at the following levels :

1. First trimester scan by 8 to 10 weeks, unless there is clear indication for still early scan study.
2. Second trimester scan around either 14 to 16 weeks or 18 to 20 weeks.
3. Third trimester scan by 32 to 34 weeks.

Need for additional sonographic studies will be decided on individualized feto-maternal conditions.

OBSERVATIONS ON FETAL ANOMALY STUDIES

Altogether 47 subjects in this study had fetal anomalies, and they could be divided into 4 groups depending on the gestational age at which the first sonographic evaluation was performed : 3 anomalous fetuses among 336 first trimester pregnancies; 8 among 170 gestations scanned between 13 and 20 weeks; 13 among 132 pregnancies scanned between 21 to 28 weeks; and 23 among 392 subjects scanned between 29 and 40 weeks (Table I). It appears that less number of fetal anomalies have been detected among those scanned in the first trimester (0.89%) as against 4.71% in the early midtrimester, and the maximum of 9.85% in the late midtrimester.

This analysis is based exclusively on the gestational age at which the patients had reported for the first ultrasound study. It goes without saying that if the same patients had been seen at an earlier period of gestation many of these anomalies would have been

Table I

Incidence of fetal anomalies diagnosed at sonography

Gestational age at which scan was done	Total pregnancies studied	No. with anomalies	%
First trimester	336	3	0.89
13 to 20 weeks	170	8	4.71
21 to 28 weeks	132	13	9.85
29 to 40 weeks	392	23	5.87

detected.

Considering only those pregnancies scanned after 12 weeks of gestation, there were 44 anomalous pregnancies for 694 subjects scanned which gives an incidence of 6.34%.

Many patients had multiple anomalies in the fetuses involving different systems, and the frequency with which various system abnormalities were located included : CNS in 27 fetuses; urinary tract abnormalities in 10 fetuses; examphalos in 9; cardiac anomalies in 8; and intestinal in 3 fetuses (Table II).

The common fetal anomalies noted in study included anencephaly (7), pen-talogy of Canter (6), hydrocephaly (5), hydronephrosis (4), Meckel Gruber syn-drome (3), encephalocele (3), diaphragmatic hernia (2), posterior urethral valve syndrome (2), and others (Table III).

Hydrocephalus and examphalos were among those anomalies diagnosed in the first trimester. Anencephaly, hydrocephalus, encephalocele, spina bifida, examphalos, ectopia cordis, renal dysplasias, and diaphragmatic hernia were the various anomalies that have been diagnosed between 13 and 20 weeks of gestation in this study. This series included all anomalies except the less common conditions like posterior urethral valve syndrome that has been diagnosed at 24 weeks, conjoined twins at 26 weeks, fetal hydrops and intestinal obstructions diagnosed in the third trimester.

A careful scrutiny revealed that a major group of anomalies which included anencephaly, encephalocele, spinal abnormality (neural tube defects), examphalos, ectopia cordis (ventral wall defects), and cystic renal dysplasia (renal anomaly) have been diag-

Table II

Frequency of fetal anomalies diagnosed in the current study

Total fetuses diagnosed to have anomalies : 47

Anomalies	Number	Percent
C. N. S. anomalies	27	57.45
Renal tract anomalies	10	21.28
Ventral abdominal wall defect	9	19.15
Cardiac anomalies	9	17.02
Gastrointestinal anomalies	3	6.38

Table III

Different types of fetal anomalies encountered in this study

Anencephaly	7	Pentalogy of Cantrel	6
Hydrocephaly	5	Encephalocele	3
Meckel Gruber syndrome	3	Diaphragmatic hernia	2
Post. urethral valves	2	Exomphalos	1
Dandy Walker Syndrome	1	Arnold Schiarri	1
Conjoined twins	1	Ileal atresia	1
Cardiomyopathy	1	Heart block	1
Thanatophoric dwarf	1	Cleft lip & palate	1
Fetal ascites	1	Fetal hydrothorax	1

Table IV

Amniotic fluid volume and fetal anomalies

Total obstetrics scans : 694	
Oligohydramnios : 45 (6.48%)	Polyhydramnios : 24 (3.46%)
Anomaly in oligohydramnios	Anomaly in polyhydramnios
14 / 45 : 31.11%	9 / 25 : 37.50%
Incidence of amniotic fluid volume abnormalities in pregnancies associated with fetal malformations	
Oligohydramnios : 14 (31.81%)	Polyhydramnios : 9 (20.45%)

nosed as early as 14 weeks. The sonographic findings in them suggested that if the same patients had reported earlier even a late first trimester diagnosis would have been made possible. Conditions like conjoined twins and fetal hydrops should not pose a problem for early diagnosis in the first trimester.

From this study we observed that only those conditions such as the intestinal obstruction, certain forms of dwarfism and cardiomyopathy could remain undetected until the third trimester.

Among those 694 subjects undergoing obstetric scans 45 had oligohydramnios and 24 polyhydramnios 6.48% and 3.46% respectively. Among the 45 subjects with oligohydramnios 14 had fetal anomalies (31.11%) and of the 24 with polyhydramnios 9 and fetal anomalies (37.50%). Of those 44 fetal anomalies located in the 2nd and third trimesters 14 had oligohydramnios and 9 had polyhydramnios, which amounts to 31.81% and 20.45% (Table IV).

DISCUSSION

By modern standards of obstetric practice where the aim is prevention and/or early detection and management of complications, the current concept of what constitutes one routine ultrasound study during the entire course of pregnancy is thoroughly inadequate. Ultrasound studied conducted at different gestational ages have different decisive roles to perform, and a single study should not be a substitute for this immense task.

First trimester scans essentially prove the intactness of the live intrauterine gravidity, and in addition gives an indirect clue to gross fetal organ development and proportionality of fetal growth. It is the early midtrimester study that is decisive in evaluation of fetal anatomy and malformations. Both these gestational ages, either individually or preferably in a complementary manner, should be ideal for determination of the gestational age. The third trimester scans exclusively aim at studying the fetal well-being, and are less suited for other indications. By the time these three studies are completed the fetal growth rate assessment, which is the hallmark in diagnosis of fetal growth abnormalities, is automatically taken care of (Rajan, 1993).

The present study overwhelmingly proves that almost all forms of fetal malformations, except conditions like gastrointesti-

nal anomalies, should remain diagnosed at an earlier period of gestation, certainly between 21 to 28 weeks. All these anomalies that have been detected late in the second trimester are amenable for an early diagnosis around 14 weeks; and majority of them even in the late first trimester. These conditions include neural tube abnormalities, ventral wall defects and renal dysplasias. Certainly the fetal skull, spine, ventral wall and limbs are clearly imaged in the late first trimester at endovaginal scan and should not pose any diagnostic problem (Rajan and Rajan, 1991). One has to be careful about physiological umbilical hernia while reported on ventral wall defects (Romero, 1991).

While the number of fetal anomalies detected during the first trimester gestation has been limited, it should be realised that all the missed abortions and growth retardations diagnosed in the first trimester represent nothing other than fetal chromosomal or structural anomalies.

By excluding fetal complications, at the first trimester and the early midtrimester scan studies, the patient is given a clear understanding that the incidence of fetal anomaly has been reduced to a negligible percentage. The subsequent studies should aim mainly at evaluating the fetal growth and well-being.

This approach to obstetric diagnosis and fetal surveillance ultimately results is elimination of abnormal pregnancies at an earlier date, and aims at optimal timing of the delivery of healthy infants, so much so the perinatal outcome is excellent with minimal perinatal morbidity and negligible mortality.

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