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ORIGINAL ARTICLE

Ultrasonographic diagnosis of fetal structural abnormalities in prenatal screening at 11–14 weeks

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PURPOSE
The aim of the study was to determine the detection rate of structural abnormalities at the 11–14 week ultrasound examination as a part of two-stage screening process in low-risk pregnancies.

MATERIALS AND METHODS
A total of 1085 consecutive low-risk pregnant women participated in screening by two-stage ultrasonography for the estimation of the sensitivity of the first and second trimester ultrasound scans in the detection of major anomalies as a part of routine screening.

RESULTS
Of 1085 pregnancies, 21 (1.93%) fetuses had at least one major structural defect considered detectable by routine ultrasound screening; 14 (1.29%) were identified at early screening and an additional 5 (0.47%) at late screening. Two abnormalities were not detected prenatally, and data were obtained from the patients after delivery.

CONCLUSION
The majority of fetal structural abnormalities can be detected by sonographic screening at 11–14 weeks, but detailed fetal anatomic survey performed at 18–22 weeks should not be abandoned.

Key words: • *pregnancy trimester, first* • *ultrasonography, prenatal* • *nuchal translucency measurement*

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Ultrasound scanning is commonly used at 11–14 weeks of pregnancy for confirmation of viability, accurate pregnancy dating, and early diagnosis of multiple gestations.

Revolutionary technological improvements and the use of high-frequency transvaginal scanning have allowed the resolution of ultrasound imaging in the first trimester to evolve to the stage where detailed early fetal development can be well visualized. Besides providing valuable information about normal embryonic development, this imaging technology has also made it possible to accurately diagnose chromosomal abnormalities and structural anomalies before the second trimester.

Until recently, attempts to detect fetal disorders of nonchromosomal origin during the first trimester have mostly been confined to high-risk groups and/or selected populations (1, 2). Although evidence is increasing that early ultrasonography in screening low-risk pregnancies for fetal structural defects might also be feasible, experiences in screening large populations are required (3–6). In this study we have evaluated detection rate of fetal structural abnormalities in routine 11–14 weeks ultrasound scanning in an unselected population.

Materials and methods

Ultrasound screening was performed at 11–14 weeks in all pregnant women who attended our ultrasound unit from 2003 to 2007. Fetal viability was examined and crown-rump length (CRL), biparietal diameter (BPD), and femur length (FL) were measured. Evaluation of fetal anatomy according to the checklist included the following: (1) Skull and brain (skull shape and cranial ossification, lateral ventricles, choroid plexus, cerebellum); (2) face (visualization of orbit and viewing fetal profile, looking for nasal bone); (3) neck (nuchal translucency [NT] measurement, search for the presence of cystic hygroma); (4) spine (examination of overlying skin and neural tube in longitudinal and transverse planes); (5) heart (heart rhythm, position, axis, four-chamber view, and examination of great vessels); (6) stomach (existence in left upper abdomen); (7) abdominal wall (examining abdominal wall and insertion of umbilical vessels); (8) kidney (existence, size and shape, tissue texture); (9) urinary bladder (existence, size, shape); (10) extremities (examining proximal and distal long bones, looking for posture of extremities). If it was not possible to clearly visualize stomach or urinary bladder at first attempt, we checked them one hour later.

Ultrasound examination was performed by the same experienced radiologist transabdominally using a 2–5 MHz probe and transvaginally using a 4–11 MHz probe (General Electric Logic5 Pro, Milwaukee, Wisconsin, USA). Fetal evaluation was performed routinely by transabdominal approach. When visualization of fetal structures was suboptimal or a

structural abnormality was suspected during transabdominal scan, transvaginal scan was always performed.

Although the present study is focused on structural abnormalities we also measured NT of the fetuses at 11 weeks–13 weeks 6 day period according to the guidelines established by Fetal Medicine Foundation (FMF) (http://www.fetalmedicine.com). We considered NT >3 mm to be increased. Increased NT not accompanied by structural defects was not included in the study, and such women were referred for counseling.

Fetal structural abnormalities classified as major and early onset were noted. Isolated choroid plexus cyst, mild ventriculomegaly, mild pelvocaliectasis, and cardiac defects not requiring treatment were not included in the statistical calculations.

Lethal, incurable, or curable severe abnormalities with a high risk of residual handicap were considered major structural abnormalities. Less severe or benign abnormalities constituted the group of minor structural abnormalities. Pregnancy outcomes were obtained from the hospital records, from clinicians, and from the patients themselves.

Results

During the 4-year period of 2003–2007, 1085 pregnant women who attended our clinic were evaluated between 11 and 14 weeks of gestational age. Forty-one cases were lost to follow-up and not seen during the second trimester. These cases were excluded from the study (according to our archives, 14 of these were lost due to miscarriage, and the remaining cases were not found recorded). The mean age of the participants at recruitment was 26 years (range, 16–46). Of the 1085 patients assessed with ultrasound examination at 11–14 weeks, 1030 (94%) had repeat ultrasound scan at 18–22 weeks, as planned.

Of 1085 fetuses, 21 (1.93%) had at least one major structural defect considered detectable by routine ultrasound screening. Fourteen (1.29%) were identified at the early screening, and an additional five (0.47%) at the late screening. Two abnormalities were not detected prenatally, and data were obtained from the patients after delivery. The mean examination time of the early scan was 13 weeks 1 day (11 weeks 5 days–14 weeks); mean examination time of the late scan was 19 weeks 2 days (18–22 weeks).

Cases detected in the first trimester included four cases of anencephaly, one encephalocele, two cases of cystic hygroma, one omphalocele with increased NT (normal karyotype), one gastroschisis, one anencephaly with spina bifida, one hydrops, one cystic hygroma with intraabdominal cyst, one holoprosencephaly (Fig. 1), and one megacystis (Table 1).

Cranial abnormalities were the most common (7/14) abnormalities detected in the first trimester. Of these, anencephaly was the most common (5/7) cranial malformation detected. Cranial abnormalities were followed by cystic hygroma (3/14), and abdominal wall defects (2/14).

All pregnancies with fetuses affected by cranial abnormalities and most of the remaining pregnancies with major malformations were terminated except two cases of cystic hygroma and one case of gastroschisis. The case with gastroschisis was operated on the neonatal period.

Five abnormalities which were not detected during routine examination at 11–14 weeks but were detected in the second trimester included three cases of hydrocephalus, one case of multicystic dysplastic kidney, and one case with cardiac anomalies.

NT ≥95th percentile for gestational age was noted in 1 of 14 (0.07%) abnormal fetuses, in which omphalocele and increased NT was detected (karyotype was normal) (Fig. 2). One of the fetuses in which the only abnormal finding was increased NT in the first trimester was counseled for karyotyping. Karyotype was normal; in the second trimester examination, a ventricu-

Table 1. Anomalies detected at the 11–14 week prenatal ultrasound scan

Anomaly	Gestational age when detected (weeks+days)	Outcome	Comments
Anencephaly	13+3	Termination of pregnancy	Malformation confirmed
Anencephaly	13+5	Termination of pregnancy	Malformation confirmed
Hydrops	11+5	Termination of pregnancy	Malformation confirmed
Omphalocele and increased nuchal translucency	13+0	Termination of pregnancy	Karyotype normal
Anencephaly and spina bifida	13+1	Termination of pregnancy	Malformation confirmed
Cystic hygroma	12+1	Live birth at week 37	Karyotype normal
Anencephaly	12+4	Termination of pregnancy	Malformation confirmed
Cystic hygroma and intraabdominal cyst	12+6	Termination of pregnancy	Malformation confirmed
Cystic hygroma	11+5	Live birth at week 37	Karyotype normal
Holoprosencephaly	13+5	Termination of pregnancy	Malformation confirmed
Anencephaly	13+3	Termination of pregnancy	Malformation confirmed
Megacystis	13+4	Termination of pregnancy	Malformation confirmed
Encephalocele	14+0	Termination of pregnancy	Malformation confirmed
Gastroschisis	13+4	Live birth at week 37	Neonatal surgery

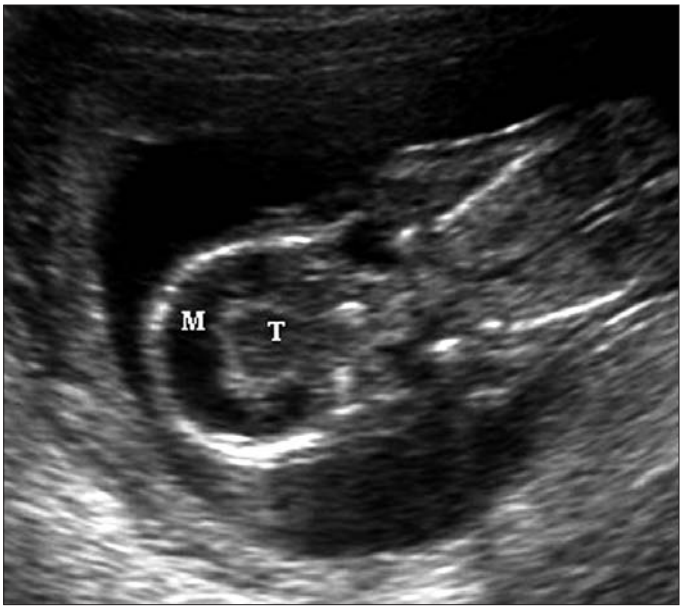

Figure 1. Coronal transabdominal ultrasound image of a fetus with holoprosencephaly at 13 weeks 5 days of gestation. Fetal head shows fused thalami (T), a monoventricle (M) with virtually no cerebral tissue, and absence of the falx and interhemispheric fissure.

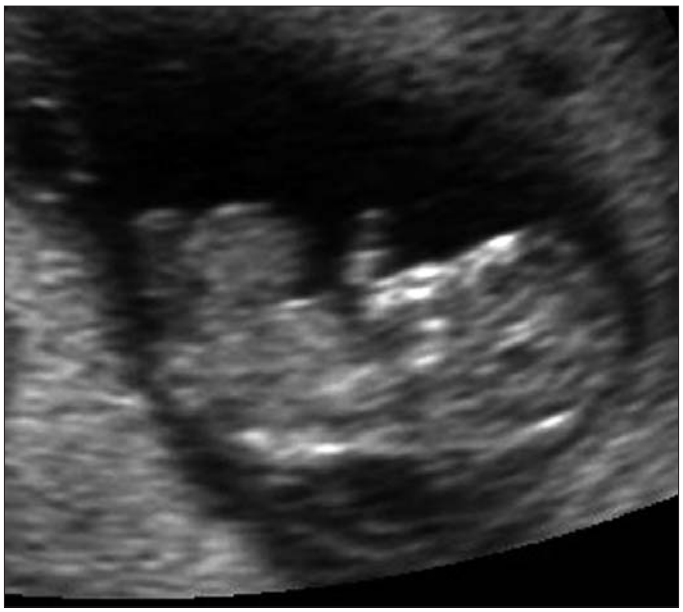

Figure 2. Sagittal ultrasound image of a fetus with omphalocele at 13 weeks of pregnancy.

lar septal defect (VSD) which was not clear in the first trimester was detected. This case was among the five cases with abnormalities that we could not detect in the first trimester but were able to detect in the second trimester examination. Two cases were not diagnosed prenatally. One neonate with pulmonary stenosis and one neonate with coarctation of the aorta were diagnosed by neonatologist postnatally.

Of 14 anomalies detected by screening at 11–14 weeks, detection rate of structural abnormalities were 50% in both the first and second years of the study, 80% and 83% in the third and fourth years of the study, respectively (Table 2).

Discussion

In this study detailed examination of fetal structures at 11–14 weeks of pregnancy revealed 66.6% of major structural abnormalities in low-risk pregnant women. Similar results have been reported from other studies (5, 7, 8), with detection rates between 22.3% and 64.7% (Table 3).

First trimester sonography is technically difficult and time-consuming, and there is a learning curve to reach an acceptable level of accuracy. In this study, when we compare the detection rates of abnormalities considering the first trimester and follow-up results in the first, second, third, and last quarters of the survey period, there was a gradual increase in success rates. To the best of our knowledge, this is the first study to compare changes in success rates in detecting abnormalities over the course of study. When we compare the first and last quarters of the study, detection rate increases from 50% to 83%, clearly emphasizing the importance of training. There are some limitations of our study which must be mentioned here. Twenty-one patients

Table 2. Detection rate of structural abnormalities at different stages of the study

Period	Anomalies detected in first trimester	Anomalies detected in second trimester	Anomalies detected postnatally	First trimester detection rate (%)	Total detection rate (%)
1st year	2	1	1	50	75
2nd year	3	2	1	50	83
3rd year	4	1	0	80	100
4th year	5	1	0	83	100

in a four-year period provided a small group for statistical analysis; studies with larger number of series must be designed to show the significance of learning curve.

A large study from Finland evaluated the learning curve over six years in detecting abnormalities in early pregnancy by transvaginal ultrasonography. It reported an increase in the detection rate from 22% to 79% over six years (9). Reevaluating the results of the screening program, the authors estimated that a two-year training program is mandatory for a sonographer to become proficient in detecting fetal anomalies (9).

In a study by Taipale et al. in 2004, only 18% of fetuses with major structural anomalies considered detectable by ultrasonography were found in early screening, and an additional 30% of such fetuses were identified at the midpregnancy scan, totaling 48% for the two-stage scan (12). These results indicated lower detection rates than reported in a study by Dane et al. (11), in which 70% of major anomalies were diagnosed at 14 weeks of gestation, and 95% when the results of a second (21-week) scan were added. The much lower detection rates obtained by Taipale et al. (12) may be partly attributable to the fact that their examiners were mostly trained midwives who used an ordinary machine and allocated about 20 minutes for each scan as part of the busy routine antenatal care. The better results obtained by Dane et al. may reflect the fact that experienced obstetricians or radiologists had a major role in the study.

Heart anomalies were not included in all the studies. Examination of the fetal heart in the first trimester was not routinely performed in the studies of Hernadi and Torocsik (5), and Carvalho et al. (10). Economides and Braithwaite included examination of the 4-chamber view of the heart, whereas in the studies of Taipale et al. (4, 9), Chen et al. (13), and Souka et al. (14) examination of the four heart chambers and the great vessels was attempted in the first trimester. In our study we routinely examined position, axis, four-chamber view, and great vessels view in all patients. First trimester echocardiography was also offered to women with increased NT.

In our study one fetus with major heart defects had NT above the 95th percentile for gestational age. The patient underwent amniocentesis, and no chromosomal abnormalities were found. This fetus had a VSD that could not be identified at the first trimester examination (12 weeks 3 days), but was correctly identified at 19 weeks of gestational age (the anomaly was confirmed by echocardiography). In comparison to other studies, we had high first trimester detection rates and overall detection rates, although we could not identify two cardiovascular anomalies. This may be partly attributable to the fact that these anomalies are rare and difficult to diagnose prenatally. Therefore we recommend cardiovascular examination by an experienced echocardiographer in patients with increased NT, even if the first trimester screening shows no abnormalities.

It is difficult to compare results from different studies because of differing definitions of malformations. There are also local differences in the prevalence of various conditions. Anomalies in our study were related predominantly to the central nervous system, with anencephaly being the most common (Fig. 3).

In expert hands, the second trimester ultrasound screening has been reported to detect up to 70% of all anomalies. Sensitivities ranging between 54% and 65% in first trimester screening have been reported in recent studies. When combined with second trimester ultrasound, sensitivities range from 77% to 82%.

Our findings are generally in accordance with previous studies. We suggest that ultrasound examination at 11 to 14 weeks to screen for fetal abnormalities is effective and can be an adjunct to the conventional examination (15). Early detection of major anomalies offers parents the option of an earlier, safer, and psychologically less traumatic termination of the pregnancy.

A detailed fetal anatomic survey performed at 18–22 weeks remains the primary method for detecting the major-

Table 3. Reported series examining the sensitivity of first and second trimester ultrasound in detecting major fetal structural defects

Authors	n	Population	Method	Major structural anomalies	First trimester detection rate (%)	Total detection rate (%)
Economides and Braithwaite	1632	Low risk	TA+TV	17 (1%)	65	82
Guariglia and Rosati	3478		TV	64 (1.8%)	52	84
Carvalho et al.	2853	Low risk	TA+TV	66 (2.3%)	38	79
Taipale et al.	4513	Low risk	TV	33 (0.7%)	18	48
Chen et al.	1609	High risk	TA+TV	26 (1.6%)	54	77
Souka et al.	1148	Low risk	TA+TV	14 (1.2%)	50	92
Cedergren and Selbing	2708	Low risk	TA	13 (1.2%)	40	
Hernadi and Torocsik	3991	Low risk		35 (0.9%)	36	72
Dane et al.	1290	Low risk	TA+TV	24 (1.86%)	70	95
Our study	1085	Low risk	TA+TV	14 (1.93%)	66	90

n, number of patients; TA, transabdominal; TV, transvaginal.

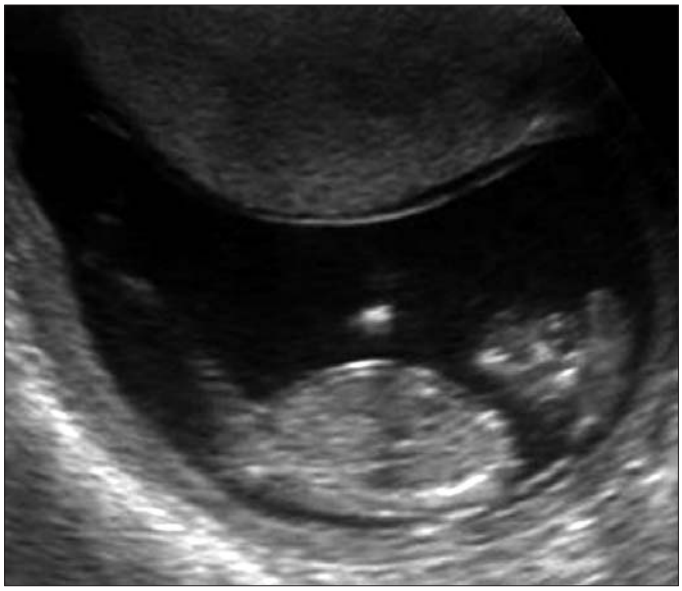

Figure 3. Sagittal ultrasound image of a 13-week-3-day fetus with absence of the cranial vault, which is consistent with anencephaly. The facial structures and orbits are present.

ity of serious structural birth defects; however, first trimester screening at 11–14 weeks has the potential to develop into the initial screening test for many patients.

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