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Effectiveness of Prenatal Ultrasonography in Detecting Fetal Anomalies and Perinatal Outcome of Anomalous Fetuses

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A retrospective study was performed over a 5-year period (1990-94) to evaluate the effectiveness of prenatal ultrasonography in terms of sensitivity, specificity, and predictive values in detecting fetal anomalies by comparing prenatal ultrasonic results with anomalies found in neonates and the perinatal outcome of anomalous fetuses. Minor congenital anomalies as listed and defined in the Eurocat Register were excluded. From a total of 5544 singletons, 4819 had at least one ultrasound scan (87%), of which 3004 at low risk and 1815 (38%) at high risk for anomalies had routine screening (RS) and indicated scanning (IS), respectively. A total of 136 fetuses were structurally abnormal (2.82%, RS and IS : 0.77% and 6.23%) and 200 major anomalies (RS and IS : 37 and 163) were recorded. The overall sensitivity of the ultrasound test was 78.7% (RS and IS : 34.8% and 87.6%, $P<0.01$) for abnormal fetuses and 58.0% (RS and IS : 29.7% and 64.4%, $P<0.01$) for anomalies. The overall specificity was 99.9% and the positive and negative predictive values were 97.3% and 99.4%, respectively; these values did not differ significantly between the two groups. The sensitivity of ultrasound for the detection of abnormal fetuses before 24 weeks was 22.8% (RS and IS : 13.0% and 24.8%) which was associated with a 61% (25/41) termination rate (RS and IS : 25% and 75.9%, $P<0.01$) and a 24.4% (10/41) postnatal survival rate (RS and IS : 41.7% and 17.2%). The overall survival rate following pre- and postnatal correction of anomalies was 44.9% (RS and IS : 60.9% and 41.6%). For the detection of fetal anomalies anatomic ultrasound scanning is necessary during pregnancy, irrespective of pregnancy condition. Early detection of fetal anomalies could offer the option of pregnancy termination.

Key Words: Ultrasound, prenatal diagnosis, fetal anomalies, perinatal outcome

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There is still much controversy over the value of routine ultrasound scanning for fetal anomalies, even though it is extensively practiced during pregnancy. European studies of routine ultrasound screening in a low-risk population have demonstrated the sensitivity of scanning to detect between 40% and 85% of fetal anomalies (Rosendahl and Kivinen, 1989; Saari-Kemppainen *et al.* 1990; Chitty *et al.* 1991; Levi *et al.* 1991; Luck, 1992; Shirley *et al.* 1992). The recent RADIUS (Routine Antenatal Diagnostic Imaging with Ultrasound) study reported that the sensitivity was as low as 35% in the detection of

fetal major anomalies (Ewigman *et al.* 1993; Crane *et al.* 1994).

When an ultrasound scan is performed on an indication basis for the detection of fetal anomalies in a high-risk population referred to specialist units, its sensitivity is much higher, ranging from 86~99% (Campbell and Pearce, 1983; Sabbagha *et al.* 1985; Manchester *et al.* 1988; Sollie *et al.* 1998).

Although the detection of fetal anomalies is enhanced by ultrasound, any beneficial effect on perinatal outcome has not yet been substantiated. The Helsinki ultrasound trial (Saari-Kemppainen *et al.* 1990) found that perinatal mortality was significantly lower in the ultrasound screened group and that a 49.2% reduction in perinatal mortality (from 9.0 per 1000 to 4.6 per 1000) was due to early detection of major malformations which led to induced abortion. This contrasts with the results of the RADIUS study (Ewigman *et al.* 1993; Crane *et al.* 1994) in which ultrasound screening did not improve perinatal outcome and had no significant impact on the frequency of abortion for fetal anomalies.

This report presents the effectiveness of prenatal ultrasonography in terms of sensitivity, specificity, and predictive values in the detection of fetal anomalies and the perinatal outcome of anomalous fetuses over the period 1990~1994 at the department of Obstetrics and Gynecology, Yongdong Severance Hospital, Yonsei University College of Medicine in Seoul, a referral hospital in which high-risk pregnancies are managed.

MATERIALS AND METHODS

A retrospective study was made to evaluate the effectiveness of ultrasound scanning in detecting fetal anomalies. From a total of 5544 singletons who were delivered at our hospital during the study period, 4819 fetuses (87%) had at least one ultrasound scan during pregnancy, of which 3004 were done on a routine basis and 1815 (38%) were referred cases on an indication basis in the form of detailed targeted imaging for fetal anomalies. Twins were excluded from this evaluation. A total of 136 fetuses were structurally abnormal at birth or at termination of pregnancy, resulting in a prevalence of major anomalies of 2.82% with multiple anomalies found in 23 fetuses (17%) (Table 1).

Routine ultrasound screening (RS) for fetal anomalies was usually performed twice during pregnancy in an unselected low-risk population, the first at 18~20 weeks and the second at 32~34 weeks of gestation. Indicated ultrasound scanning (IS) was determined on a selective basis in high-risk pregnancies at risk for birth defects, which usually included a family history of congenital anomalies, advanced maternal age (≥35 years), either high or low levels of maternal serum alpha-fetoprotein, either small or large uterine fundal size, exposure to teratogens and suspicious outside ultrasound findings. Scans were obtained with Acuson (model 128) or Ultramark (model 9) scanners by a trained obstetric fellow.

Anatomical scanning of the fetus was done as recommended by Campbell and Pearce (1983). When a fetal anomaly is detected or suspected, the mother is referred for further detailed scanning by a senior obstetric consultant (K. Lee). If an anomaly is found, parents are counselled as to the findings and further investigations are undertaken such as amniocentesis or cordocentesis for karyotyping in certain

Table 1. Characteristics of study populations

Singleton pregnancies		Fetuses scanned	%	Indicated scan %	Abnormal fetuses with anomalies			
					single	multiple	total	%
1990	822	674	82	18	15	2	17	2.52
1991	976	803	82	27	18	6	24	2.99
1992	1209	1212	84	36	23	4	27	2.67
1993	1259	1119	89	51	34	5	39	3.49
1994	1278	1211	95	45	23	6	29	2.39
Total	5544	4819	87	38	113	23	136	2.82

anomalies, management and prognosis.

Since April 1983 we have kept a detailed record of all anomalies detected by ultrasound. We reviewed this record for 1990～1994 and confirmed the pregnancy outcomes by reviewing the maternal and neonatal notes. Sources of ascertaining fetal anomalies included clinical newborn examinations, 58 autopsy findings out of 73 deaths, 50 chromosomal analyses, radiologic studies and 25 operative findings among 63 neonates. The records of infants born during the study period were also checked from 2 months to 2 years after birth to detect any anomalies missed in scanning.

A congenital anomaly is an anatomical or structural abnormality present at birth, including malformation, deformation, disruption and dysplasia (Spranger *et al.* 1982). Fetal anomalies were classified according to the International Classification of Disease 10 (1992) and included nonimmune hydrops fetalis and tumor. For this study we excluded the minor anomalies defined by Smith (1982) and the Eurocat Register (De Wals *et al.* 1984).

The sensitivity, specificity, and predictive values of both RS and IS in detecting fetal anomalies were calculated by comparing prenatal ultrasonic results with anomalies found in neonates. Perinatal outcome was defined as termination of pregnancy, fetal death, neonatal death and live birth. For statistical analysis the chi-square test with Yate's correction or Fisher's exact test was used.

RESULTS

A total of 136 fetuses (2.82%) were structurally abnormal at birth or at termination of pregnancy ; 23 and 113 fetuses were respectively in the RS and IS groups. Of the 136 fetuses with major anomalies, 107 were detected by ultrasound with an overall sensitivity of 78.7%. The respective figures for sensitivity were significantly different ($P<0.01$) for RS (34.8%) and for IS (87.6%). Prenatal detection of abnormal fetuses was correctly made before 24 weeks gestation in 31 cases (22.8%) and early detection was higher in IS (28/113 or 24.8%) than in RS (3/23 or 13.0%) (Table 2).

As predicted, 4680 fetuses were normal at birth or on discharge from hospital, giving a specificity of 99.94% and this was not significantly different between RS (100.0%) and IS (99.82%). Three fetuses were erroneously suspected of having a defect, the false-positive rate being 0.06%. The 3 ultrasound diagnoses that were not confirmed by postnatal examination were ovarian cyst, heterozygous achondroplasia and esophageal atresia, respectively in IS.

Table 2. Routine and indicated ultrasound results

	Routine	Indicated	Total
Fetuses scanned	3004	1815	4819
Abnormal fetuses	23	113	136
True positives	8	99	107
<24 weeks	3	28	31
≥24 weeks	5	71	76
True negatives	2981	1699	4680
False positives	0	3	3
False negatives	15	14	29
Sensitivity(%)	34.8	87.6	78.7
<24 weeks	13.0	24.8	22.8
Specificity(%)	100.0	99.82	99.94
Predictive value(%)			
positive	100.0	97.06	97.27
negative	99.5	99.18	99.38
Anomalies	37	163	200
Anomalies detected	11(29.7%)	105(64.4%)	116(58.0%)
<24 weeks	5(13.5%)	33(20.2%)	38(19.0%)
Anomalies not detected	26	58	84

None of these false-positive diagnoses were followed by termination. The positive predictive value was 97.27% (RS and IS : 100.0% and 97.06%) and the negative predictive value was 99.38% (RS and IS : 99.5% and 99.18%)

The 136 abnormal fetuses had 200 major anomalies. Considering the number of anomalies, the sensitivity was found to be 58.0% (116/200), and there was a significant difference ($P<0.01$) between RS (29.7% or 11/37) and IS (64.4% or 105/163). Ultrasound correctly detected anomalies before 24 weeks in 38 (19.0%) of the 200 anomalies: RS 5 (13.5%) and IS 33 (20.2%). The highest sensitivities were obtained in the detection of central nervous system abnormalities in RS and IS, 66.7% and 77.4% respectively and abnormalities of the genitourinary tract, 50.0% and 77.4%. The lowest sensitivities of IS were obtained for the face: 22.2%, skeletal abnormalities: 31.3%, and cardiovascular abnormalities: 40.0% (Table 3a and 3b). In Table 4, the details of anomalies are displayed for each system in relation to the number of anomalies, the number detected before 24 weeks and after 24 weeks and the false negatives.

Fifty fetuses had karyotype determination (15 by amniocentesis, 12 by cordocentesis, 2 by cardiocentesis followed by fetocide, 21 by postnatal umbilical cord blood sampling and skin biopsy) after a structural abnormality had been identified. Of the 12 fetuses with an abnormal karyotype, 4 had been

Table 3a. Sensitivity of prenatal routine ultrasound in detecting major anomalies displayed by individual systems

Defects	Total N	Prenatal detection N	Prenatal detection %	Early(<24 weeks) Prenatal detection N	Early(<24 weeks) Prenatal detection %
Gastrointestinal tract and wall	6	2	33.3	1	16.7
Central nervous system	3	2	66.7	1	33.3
Urogenital system	2	1	50.0	0	0.0
Cardiovascular system	6	2	33.3	0	0.0
Skeleton and limbs	4	0	0.0	0	0.0
Face	8	0	0.0	0	0.0
Hydrothorax/Ascites/Hydrops	2	2	100.0	2	100.0
Chromosome	5	1	20.0	0	0.0
Neck	1	1	100.0	1	100.0
Total	37	11	29.7	5	13.5

Table 3b. Sensitivity of prenatal indicated ultrasound in detecting major anomalies displayed by individual systems

Defects	Total N	Prenatal detection N	Prenatal detection %	Early(<24 weeks) Prenatal detection N	Early(<24 weeks) Prenatal detection %
Gastrointestinal tract and wall	29	18	62.1	3	10.3
Central nervous system	31	24	77.4	11	35.5
Urogenital system	31	24	77.4	7	22.6
Cardiovascular system	15	6	40.0	1	6.7
Skeleton and limbs	16	5	31.3	2	12.5
Face	9	2	22.2	0	0.0
Hydrothorax/Ascites/Hydrops	14	13	92.9	4	28.6
Chromosome	7	3	42.9	2	28.6
Neck	5	4	80.0	3	60.0
Lung	3	3	100.0	0	0.0
Others	3	3	100.0	0	0.0
Total	163	105	64.4	33	20.2

detected to have a structural abnormality. Eight had not been detected: 5 Trisomy 21 and 1 mosaicism (47, XX, +21/46, XX) had no identifiable structural abnormalities. If the 6 cases with no associated structural abnormality are deducted from the 136 abnormal fetuses, sensitivity of detection of abnormality becomes 82.3% (107 of 130).

Perinatal outcome of 136 abnormal fetuses is presented in Table 5. Pregnancy terminations, fetal deaths, neonatal deaths and survivals following pre- and postnatal correction of their congenital anomalies occurred in 39%, 5.9%, 8.8% and 44.9%, respectively.

Detection of anomalous fetuses before 24 weeks was associated with a 61% (25/41) termination rate (RS and IS : 25% and 75.9%, $P<0.01$) and a 24.4%

Table 4. Details of major anomalies detected in the indicated and (routine) ultrasound scans

	Total number of anomalies	True positives			False negatives
			Age of detection		
		n	<24 weeks	≥24 weeks	n
Central nervous system					
Anencephaly	15	15	7	8	0
Encephalocele			0		0
Spina bifida	3		-	0	2
Hydrocephalus	7	6	2	4	1
Microcephaly	3	0	0	0	2(1)
Holoprosencephaly	2	(1)	0	(1)	1
Ventriculomegaly	1	(1)	(1)	0	0
Dandy-Walker malformation	-	-	-	0	0
Agenesis of corpus callosum		0	0	0	1
Total	34	24(2)	11(1)	13(1)	7(1)
Cardiovascular system					
Ventricular septal defect	7	2(2)	0	2(2)	1(2)
Atrial septal defect	3	1	0	1	2
Atrioventricular septal defect	2	-	0	1	-
Aortic stenosis	1	0	0	0	(1)
Single ventricle	1	0	0	0	1
Tetralogy of Fallot	1	0	0	0	(1)
Hypoplastic left heart syndrome	1	0	0	0	-
Ectopia cordis	-	-	-	0	0
Situs inversus	4		0	-	3
Total	21	6(2)	1	5(2)	9(4)
Gastrointestinal tract and wall defects					
Esophageal atresia	4	1	0		2(1)
Duodenal atresia	3	3	0	3	0
Jejuno-ileal atresia	4	3	0	3	(1)
Multiple atresia	-	0	0	0	
Anorectal atresia*	4	0	0	0	3(1)
Diaphragmatic hernia	5	1(1)	0	1(1)	2(1)
Omphalocele	6	3(1)	1(1)	2	2
Gastroschisis	2	2	1	-	0
Body stalk anomaly	1	0	0	0	1
Abdominal wall defect	-	-	1	0	0
Meconium peritonitis/pseudocyst	3	3	0	3	0
Choledochal cyst	1	1	0	-	0
Total	35	18(2)	3(1)	15(1)	11(4)

Table 4. Continued

	Total number of anomalies	True positives			False negatives
			Age of detection		
		n	<24 weeks	≥24 weeks	n
Urogenital system					
Multicystic kidney	7	6	3	3	
Renal cystic dysplasia	2		0	1	1
Hydronephrosis	6	6	0	6	0
Ureteropelvic junction obstruction	4	3	2	1	1
Urethral atresia	1	1		0	0
Vesicoureteral reflux	1	0	0	0	1
Absence of bladder	1	0	0	0	1
Prune-belly syndrome	1	1		0	0
Horseshoe kidney	2	0	0	0	1(1)
Single discoid kidney	1	0	0	0	
Mesoblastic nephroma	1	1	0		0
Ovarian cyst	5	4(1)	0	4(1)	0
Parovarian cyst		1	0		0
Total	33	24(1)	7	17(1)	7(1)
Skeleton and limbs					
Thanatophoric dysplasia	2	2	0	2	0
Craniosynostosis*	1	0	0	0	1
Cranial defect	1	0	0	0	1
Absent lumbosacral spine			1	0	0
Absent left radius		0	0	0	1
Failure of development of right arm		0	0	0	1
Right humeroradial synostosis*	1	0	0	0	1
Absent left leg	1		1	0	0
Phocomelia of right leg	1	0	0	0	1
Absent sternum	1	0	0	0	1
Absent clavicles	1	0	0	0	
Clubfoot and clubhand	3	1	0		(2)
Rockerbottom foot	2	0	0	0	(2)
Amputation of fingers	1	0	0	0	1
Amputation of toes	1	0	0	0	
Absent distal phalanx of both big toes	1	0	0	0	
Total	20	5	2	3	11(4)
FACE					
Cleft lip, cleft palate,* or both	15	2	0	2	7(6)
Cyclopia	1	0	0	0	0(1)
Proboscis	1	0	0	0	0(1)
Total	17	2	0	2	7(8)
Nonimmune hydrops fetalis	11	8(2)	3(2)	5	
Isolated hydrothorax	3	3	0	3	0
Isolated ascites	2	2	1	1	0
Total	16	13(2)	4(2)	9	1
Chromosomal abnormality					
Trisomy 21	6	1	0	1	2(3)
Turner syndrome	2	2	2	0	0
Trisomy 18	3	(1)	0	(1)	1(1)
47, XX, +21/46, XX	1	0	0	0	1
Total	12	3(1)	2	1(1)	4(4)

Table 4. Continued

	Total number of anomalies	True positives			False negatives
			Age of detection		
		n	<24 weeks	≥24 weeks	n
Neck					
Cystic hygroma	6	4(1)	3(1)		1
Lung					
Congenital cystic adenomatoid malformation of lung	3	3	0	3	0
Others					
Retroperitoneal teratoma	1	1	0	1	0
Sacrococcygeal teratoma	2	2	0	2	0
Total	3	3	0	3	0
Grand total	200	105(11)	33(5)	72(6)	58(26)

*: Anomaly not detectable by ultrasound

Table 5. Perinatal outcome of 136 abnormal fetuses

	Routine		Indicated		Total (%)
	<24 weeks	≥24 weeks	<24 weeks	≥24 weeks	
Termination of pregnancy	3	0	22	28	53(39.0)
Fetal death	1	1	2	4	8(5.9)
Neonatal death	3	0	0	9	12(8.8)
Alive	5	9	5	42	61(44.9)
Discharged against advice	0	1	0	1	2(1.5)
Total	12	11	29	84	136(100.0)

(10/41) postnatal survival rate (RS and IS : 41.7% and 17.2%), whereas detection at 24 weeks or more was associated with a 29.5% (28/95) termination rate (RS and IS : 0% and 33.3%) and a 53.4% (51/95) postnatal survival rate (RS and IS : 81.8% and 50%).

The survival rate was 44.9% for all the anomalous fetuses; 60.9% (14/23) in the RS group and 41.6% (47/113) in the IS group. In this study, 6 babies had fetal surgery with a good outcome in 2; one open surgery for diaphragmatic hernia and 5 shunts for 2 hydrothoraces, one isolated ascites, one urethral atresia and one prune-belly syndrome, respectively. Twenty-five babies among 63 neonates had early neonatal surgery with a good outcome in 23. In 18 of these 25 cases, the diagnosis had been accurately made by prenatal ultrasound. This suggests that there is the potential for perinatal mortality to be further reduced by fetal surgery when abnormality has been detected prenatally.

DISCUSSION

In this study prenatal ultrasound had a detection rate of 78.7% of abnormal fetuses. The detection rate of 34.8% in the RS group was significantly

lower than the rate in the IS group (84.6%).

Our sensitivity rate for RS was similar to that (34.8～36%) reported by the RADIUS study group (Ewigman *et al.* 1993; Crane *et al.* 1994) and Goncalves *et al.* (1994), but lower than that (40.4～58.1%) reported in other studies(Rosendahl and Kivinen, 1989; Luck, 1992). Our rate of sensitivity for IS was similar to that (86%) reported by Sollie *et al* (1998). but lower than that (95～99%) reported in other studies (Campbell and Pearce, 1983; Sabbagha *et al.* 1985; Manchester *et al.* 1988). This result does not indicate that IS rather than RS should be performed on a selective basis for the detection of fetal anomalies. This is because 90% of infants with congenital anomalies and chromosome abnormalities are born to healthy young women with no identifiable risk factors (Royal College of Physicians, 1989).

The sensitivity of ultrasound in the detection of fetal anomalies is dependent on the prevalence of anomalies in a study population, the expertise of the examiner, the gestational age at scanning, the definition of anomaly-major and minor, and the postnatal ascertainment of anomalies.

The prevalence of major fetal anomalies (2.82%) in this study was within the range of 2～3% reported in some epidemiological studies (Marden *et al.* 1964; Ekelund *et al.* 1970), although in the RS and IS groups, the respective prevalence was lower [0.77% (23/3004) : 6.23% (113/1815)] than that (0.99～2.45%) reported in the studies on RS(Chitty, 1995) and that (13.6～21.0%) on IS (Campbell and Pearce, 1983; Sabbagha *et al.* 1985; Sollie *et al.* 1998). When high-risk populations are studied, the prevalence of fetal anomalies are much higher.

The skill and experience of the sonographers is a critical factor in the detection of fetal anomalies. The detection rates were better in teaching hospitals with tertiary referral units than in local units (13% : 35% Ewigman *et al.* (1993); 36% : 76.9% Sarri-Kemppainen *et al.* (1990)). Obstetricians in private offices detected 22%, the examiner in the hospital 40%, and the examiner in the centre for prenatal diagnosis and therapy 90% of all fetal malformations (Bernaschek *et al.* 1996). The recent review of the routine screening program showed improved early detection rates from 21% in the period 1984～89 to 41% in 1990～92 (Levi *et al.* 1995). Much of this improvement can be attributed to increased experience and training. With the involvement of specialists working full-time in the field of prenatal sonographic diagnosis during the later study periods (1990～91), the sensitivity of ultrasound rose to 96% (Carrera *et al.* 1995).

There is considerable variation in the sensitivity of ultrasound scannings in the detection of anomalies in different systems. The lowest sensitivities were found for facial, limb, skeletal and cardiovascular anomalies, as in other studies (Rosendahl and Kivinen, 1989; Chitty *et al.* 1991; Levi *et al.* 1991; Shirley *et al.* 1992; Goncalves *et al.* 1994; Levi *et al.* 1995). Our data support the necessity of continual improvement in obstetric ultrasonographic services. We suggest that views of the fetal face and cardiac outflow tracts as well as the 4-chamber should be an element of routine obstetric ultrasonographic examination.

Second-trimester RS in European centers for the detection of fetal anomalies provides a sensitivity rate of 50.9% (Romero, 1993) and ranges from 21% to 85% (Rosendahl and Kivinen, 1989; Saari-Kemppainen *et al.* 1990; Chitty *et al.* 1991; Levi *et al.* 1991; Luck, 1992; Shirley *et al.* 1992). The earlier screening between 12/15 and 22 weeks in Belgium (Levi *et al.* 1991) and the RADIUS study (Crane *et al.* 1994) may account in part for the lower detection rates of 21% and 17%, respectively. Better detection rates (60.7%～85%) are reported from the UK (Chitty *et al.* 1991; Luck, 1992; Shirley *et al.* 1992) where screening is performed at 18～20 weeks. When additional scans were carried out in the third trimester to rule out late onset (e.g. hydrocephalus, hydronephrosis, intestinal atresia) and late manifestation (e.g. absence of corpus callosum, achondroplasia, microcephaly) anomalies, the sensitivity rates of detection increased more than twice in both RS (Rosendahl and Kivinen, 1989; Levi *et al.* 1991; Crane *et al.* 1994) and IS (Hegge *et al.* 1989; Goncalves *et al.* 1994) as in our study. Recent endovaginal ultrasound in the first trimester allows a high frequency of detection of fetal anomalies (Cullen *et al.* 1990; Rottem and Bronshtein, 1990; Achiron and Tadmor, 1991; Harrington *et al.* 1993).

The definition of anomalies included in the reported studies is variable. Some authors included anomalies such as patent ductus arteriosus (Crane *et*

al. 1994; Levi *et al.* 1995), hypospadias (Goncalves *et al.* 1994), congenital dislocation of hip (Crane *et al.* 1994; Goncalves *et al.* 1994) and umbilical artery absence (Goncalves *et al.* 1994) or excluded chromosomal abnormalities without the associated structural anomalies (Chitty *et al.* 1991; Luck, 1992) and facial clefts (Luck, 1992) in contrast with our selection criteria of major anomalies. Chromosomal anomalies per se are not detectable by ultrasound. When chromosomal abnormalities are strongly suggested by ultrasound, it is through the visualization of structural anomalies indicating the need for karyotyping. The sensitivity of ultrasound in detecting fetal chromosomal abnormalities in low-risk populations is approximately 34~35% (Stoll *et al.* 1993). It is crucial to clearly define major and minor anomalies in the evaluation of the diagnostic accuracy of ultrasound scanning for fetal anomalies.

Skupski *et al.* emphasized the use of the same outcome: major anomalies detectable by ultrasonography for the comparison of studies examining the effectiveness of prenatal ultrasound screening for congenital anomalies (Skupski *et al.* 1996). In this study there were 16 major anomalies and 9 abnormal fetuses not detectable by ultrasonography; 4 anorectal atresia (2 isolated imperforate anus, 1 imperforate anus associated with cleft palate, and 1 caudal regression syndrome including absence of rectum), one each of craniosynostosis and humeroradial synostosis in Antley-Bixler syndrome and 10 cleft palates (5 isolated, 3 associated with cleft lip, 1 associated with imperforate anus already included in the anorectal atresia, and one amniotic band syndrome including cleft lip and palate). If major anomalies detectable by ultrasonography are included, our sensitivity in detecting anomalies and abnormal fetuses rises to 63.0% (116/184) and 84.3% (107/127), respectively. If we further exclude 6 cases of chromosomal abnormalies not associated with structural anomaly, the sensitivity of abnormal fetuses becomes 88.4% (107/121).

Complete postnatal ascertainment of anomalies with the follow-up period may account to some degree for the apparently poor performance of the scanning reported from Beligum (Levi *et al.* 1991; Levi *et al.* 1995) and RADIUS (Ewigman *et al.* 1993; Crane *et al.* 1994).

Routine and indication-based screening for fetal malformations has been compared (Bernaschek *et al.* 1994). More malformations were detected before 24 weeks by means of RS (18%) than IS (5%). This result was opposed to our results that more malformations were detected on IS. This was a retrospective study comparing the different policies in operation at different times, IS in 1983~84 and RS in 1990~91. Therefore the improved technology and understanding between the two study periods may have influenced these results. Hegge *et al.* have emphasized that an indication-based system of referral for obstetric ultrasound not only fails to detect abnormal fetuses in pregnancies without indications for ultrasound, but also fails to detect abnormal fetuses sufficiently early in pregnancies with indications to permit the entire spectrum of managment choices, including termination (Hegge *et al.* 1989).

The specificity reported for most mid-trimester ultrasound screening programs is high (99.9~100%) (Rosendahl and Kivinen, 1989; Saari-Kemppainen *et al.* 1990; Chitty *et al.* 1991; Levi *et al.* 1991; Luck, 1992; Shirley *et al.* 1992) and thus the majority of parents are correctly assured. However, a few parents will be falsely assured and their babies will have an unexpected abnormality. The false-positive rate of diagnostic ultrasound is extremely low (less than 2%) in experienced hands (Romero, 1993).

Major structural anomalies account for 20-to-30% of perinatal deaths (Morrison, 1985). In this study period a total of 203 perinatal deaths occurred, of which 65 (32%) resulted from major anomalies. The outcome of anomalous fetuses depends on the gestational age at the detection of anomalies, the severity of the congenital anomalies diagnosed, the parents' decision regarding termination of pregnancy and the availability of effective treatments, either pre-or postnatally.

Of all the screened pregnancies, the incidence of elective abortion for fetal anomalies detected by midtrimester RS has been reported from 0.1% to 0.6% (Chitty, 1995). In this study the rate of pregnancy terminations before 24 weeks after the diagnosis of major fetal anomalies was 25% in the RS group, while it was 21% in the RADIUS study (Ewigman *et al.* 1993) and 37% in the Helsinki trial (Saari- Kemppainen *et al.* 1990). In the IS group, the termination rate was 76%, about 3 times higher than that (25%) in the RS group ($P<0.01$). Overall,

our pregnancy termination rate (44%) of anomalous fetuses in the IS group was significantly higher than that (13%) in the RS group ($P<0.05$). Manchester *et al.* reported a termination rate of 18% in pregnancies complicated by suspected fetal anomalies (Manchester *et al. 1988).* Early detection of anomalies was found to be associated with a increased termination rates; IS detection of abnormal fetuses at 22 weeks or less was associated with a 67% termination rate and an 11% postnatal survival rate, whereas detection at 23 weeks or more was associated with a 14% termination rate and a 51% postnatal survival rate (Rottem and Bronshtein, 1990).

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