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## The performance of routine ultrasonographic screening of pregnancies in the Eurofetus Study

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### Abstract

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**Objectives:** The purpose of the Eurofetus Study was to evaluate the accuracy of the antenatal detection of malformations by routine ultrasonographic examination in unselected populations.

**Study Design:** All ultrasonographic diagnoses of malformations and the outcomes of the fetuses were prospectively recorded in 61 European obstetric units over a 3-year period (1990-1993). Also recorded were all cases of malformation diagnosed after abortion or birth for the mothers who underwent follow-up in these centers.

**Results:** Of 3685 malformed fetuses, 2262 had received diagnoses during pregnancy (sensitivity, 61.4%). Of a total number of 4615 malformations, 2593 were detected (sensitivity, 56.2%). The detection sensitivity was higher for the major than for the minor abnormalities (73.7% vs 45.7%), and the diagnosis was made earlier in the pregnancy (24.2 weeks vs 27.6,  $P < .01$ ). Overall, 55% of the major abnormalities were detected within 24 gestational weeks. Within each severity group the accuracy of detection depended on the system. For the major abnormalities it was better for the central nervous system (88.3%) and urinary tract (84.8%) but lower for the heart and great vessels (38.8%). Detection of minor abnormalities was also effective for the urinary tract (89.1%) but not for the heart and great vessels (20.8%) or the musculoskeletal system (18%). Detection of abnormalities had an influence on the rate of termination of pregnancy. The rate of live births for the mothers bearing fetuses with major abnormalities was lower than that for the mothers in whom no abnormalities were detected, mainly because of the higher rate of elective terminations of pregnancy in the former group.

**Conclusion:** Systematic ultrasonographic screening during pregnancy can now detect a large proportion of fetal malformations, although some still escape detection. (*Am J Obstet Gynecol* 1999;181:446-54.)

(Click on a term to search this journal for other articles containing that term.)

**Key words:** [Ultrasonography](#), [screening](#), [fetal anomalies](#)

The last 2 decades have seen considerable advances in obstetric ultrasonography, and this examination now forms part of routine antenatal care in most European countries. It can determine the gestational age, detect multiple pregnancies, evaluate fetal growth, and detect malformations. Advances in imaging have led to improvements in the assessment of fetal development and identification of malformations.

Despite their relatively low prevalence (ranging from 2% to 4% of all births), fetal malformations are responsible for approximately 30% of perinatal deaths in developed countries. Diagnosis of malformation provides information for decisions during pregnancy, appropriate treatment at birth, and prompt transfer to units specializing in the care of the newborn. Therefore prenatal detection of malformations should help reduce perinatal mortality and morbidity, and routine ultrasonographic screening appears to be well justified.

However, the reliability and value of screening for congenital malformations by routine ultrasonography have been the subject of debate in recent years, especially in light of studies with contradictory findings.<sup>1,2</sup>

A considerable range in the detection rate (14% to 90%) for malformations has been reported.<sup>1,3-6</sup> Most of the studies have been retrospective or have analyzed a limited series of malformations, largely because they are rare. However, because of the diversity of malformations, the overall reliability of screening can be evaluated only in a large population. The Eurofetus Study was designed to fulfill this objective.

### Material and methods

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### Study design.

During the study period January 1, 1990–June 30, 1993, the 61 centers participating in the Multicentric Eurofetus Study recorded prospectively all diagnoses of malformation made during pregnancy by ultrasonographic examination. Also included were the defects found at birth that had not been identified despite examination carried out in the participating centers. These centers were distributed throughout 14 countries in Europe (Austria, Belgium, Croatia, Finland, France, Great Britain, Hungary, Italy, Luxembourg, Norway, Poland, Portugal, Spain, and Sweden). The main recommendation for screening was at least 1 systematic examination of the fetus, preferably between 18 and 22 weeks' gestational age. All centers performed routine ultrasonography during pregnancy and were able to record all cases of malformations after birth or abortion. Ultrasonographic examinations were carried out by qualified personnel with high-quality equipment and could be considered as "level 2."

Because the main goal of the study was to assess the reliability of routine ultrasonographic screening, any pregnant women referred for suspected malformation on ultrasonographic examinations performed outside these centers were excluded.

#### Data collection.

Two sets of information were recorded in this prospective survey. (1) For the malformations detected by ultrasonographic examination during pregnancy, data were recorded at 2 different times. The nature of the malformation, any associated signs, and decisions based on this diagnosis (further examinations and pregnancy and management) were recorded. After birth or abortion, the following information was recorded: confirmation or exclusion of the abnormalities initially recorded, course of pregnancy, follow-up after initial diagnosis (further investigations and examinations and management), outcome of pregnancy, mode of termination (birth or abortion), the malformations observed, and the status of the infant at birth and at age 6 days. (2) The malformations discovered after abortion or within 6 days after birth were recorded for fetuses or infants whose mothers had at least 1 ultrasonographic examination performed in the center. The details of the abnormalities, the dates of the ultrasonographic examinations carried out during pregnancy, the outcome of the pregnancy, and the status of the infant were recorded.

#### Definition of cases.

We defined congenital abnormalities as the various structural defects noted at birth, which include malformations, deformations, and dysplasias. We excluded the abnormalities without serious medical consequences (minor deformities of the nose, ears and face, clicking hip, umbilical or inguinal hernia, undescended testes, hydrocele, phimosis, hypospadias, isolated skin lesions, functional cardiac murmurs, and isolated single umbilical artery). Isolated growth restriction, amniotic fluid abnormalities, hydrops, and choroid plexus cysts were also excluded.

Chromosomal abnormalities in the absence of structural deformities were also recorded but are not presented in this article.

#### Data analysis.

The data during pregnancy from each patient were compared with those obtained at birth or those obtained on autopsy (for the elective terminations and perinatal deaths). An ultrasonographic diagnosis was classified as (1) true positive when it was confirmed by examination at term (clinical, paraclinical, or autopsy findings), (2) false negative if it was observed at term but was not detected on ultrasonographic examination during pregnancy, (3) false positive if it was recorded before birth but was not confirmed by examinations at term, or (4) false alarm if an abnormality was suspected initially but was not confirmed on subsequent ultrasonographic examinations. These were in fact "true negatives."

We distinguished major malformations from minor ones. Major malformations were lethal abnormalities or those that were incurable and liable to incur marked handicap or those requiring major surgical intervention. The remainder were classified as minor malformations. The latter thus formed a heterogeneous group of less severe or benign abnormalities.

The statistical analysis was performed on 2 units: the malformed fetus and the malformation itself. The malformations and their coding were based on the *International Classification of Diseases, Ninth Revision (ICD-9)*.

Results

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#### Abnormalities observed.

During the study period 4615 malformations were recorded in 3686 fetuses, representing 1.25 abnormalities per malformed fetus. Of these 3686 malformed fetuses, 2907 (78.9%) had a single malformation, 484 (13.1%) had 2 malformations, and 295 (8.0%) had  $\geq 3$  malformations (including ICD-9 code 7597). In addition, 266 malformed fetuses had a chromosomal abnormality that was not recorded as an additional malformation.

For the total study population, the most common malformations were those of the musculoskeletal system (22.6%), urinary tract (20.7%), heart and large vessels (20.7%), and central nervous system (16.0%).

#### Sensitivity of screening.

The overall sensitivity of ultrasonographic screening for a malformed fetus was 61.4%. It was significantly higher for the fetuses with several malformations than for those with a single malformation (odds ratio, 1.77; 95% confidence interval, 1.49-2.11).

Of the 4615 malformations recorded in our series, 2593 were diagnosed during pregnancy. The overall sensitivity for detection of malformations by ultrasonography in this unselected population was 56.2% (95% confidence interval, 54.7-57.7).

Considering the major abnormalities, the overall sensitivity of detection was 73.7% versus only 45.7% for the minor abnormalities.

It can be seen from the results shown in Table I that there were marked differences in sensitivity for the different malformations.

**Table I.** Detection sensitivity as function of malformations

Anomaly	ICD-9 code	No.	True positive (No.)	Sensitivity (%)
Central nervous system anomalies				
Anencephaly	7400	157	156	99.4
Cranioschisis	7401	3	3	—
Spina bifida and hydrocephalus	7410	92	87	94.6

Spina bifida without hydrocephalus	7419	89	59	66.3
Encephalocele	7420	48	41	85.4
Microcephalus	7421	38	26	68.4
Reduction deformities of brain	7422	37	32	86.5
Hydrocephalus	7423	201	188	93.5
Other specified anomalies of brain	7424	65	53	81.5
Other specified anomalies of spinal cord	7425	2	1	—
Unspecified anomalies of central nervous system	7429	6	6	—
TOTAL central nervous system anomalies	—	738	652	88.3
Conjoined twins	7594	5	4	—
Multiple congenital anomalies	7597; 7598	115	86	74.8
Heart and great vessel anomalies				
Major anomalies				
Common truncus	7450	22	13	59.1
Transposition of great vessels	7451	66	14	21.2
Tetralogy of Fallot	7452	49	24	49.0
Common ventricle	7453	22	10	45.5
Endocardial cushion defect	7456	48	28	58.3
Cor biloculare	7457	1	0	—
Other bulbus cordis	7458	2	2	—
Anomalies of pulmonary valve	7460	32	7	21.9
Tricuspid atresia and stenosis	7461	11	6	—
Ebstein's anomaly	7462	4	1	—
Stenosis of aortic valve	7463	16	2	—
Hypoplastic left heart syndrome	7467	44	24	54.5
Coarctation of aorta	7471	26	1	3.8
Other anomalies of aorta	7472	10	0	—
Other specified severe anomalies of heart	7468	13	10	—
Total major anomalies of heart	—	366	142	38.8
Minor anomalies				
Ventricular septal defect	7454	365	42	11.5
Atrial septal defect	7455	97	13	13.4
Unspecified defect of septal closure	7459	3	0	—
Mitral stenosis	7465	2	1	—
Mitral insufficiency	7466	3	1	—
Other specified minor anomalies	7468	47	34	71.7
Unspecified anomalies of heart	7469	35	18	51.4
Anomalies of pulmonary artery	7473	19	7	—
Anomalies of great veins	7474	6	0	—
Other specified anomalies of circulatory system	7478	1	1	—
Situs inversus	7593	9	5	—
Total minor anomalies of heart	—	587	122	20.8
TOTAL heart and great vessel anomalies	—	953	264	27.7
Digestive system anomalies				
Occlusion	7512	8	8	—
Minor anomalies				
Tongue anomalies	7501	3	0	—
Esophageal atresia and stenosis	7503	58	29	50.0
Other specified anomalies of stomach	7505; 7507	4	2	—
Meckel's diverticulum	7510	2	1	—
Atresia of small intestine	7511	69	60	87.0
Atresia of anus and rectum	7512	52	8	15.4
Congenital distention of colon	7513	3	2	—
Anomalies of intestine fixation	7514	4	1	—

Other anomalies of intestine	7515	6	2	—
Anomaly of gallbladder, bile ducts, liver	7516	12	7	—
Other anomalies of intestine	7518; 7519	8	3	—
Total minor anomalies of digestive system	—	221	115	52.0
TOTAL digestive system anomalies	—	229	123	53.7
Urinary tract anomalies				
Major anomalies				
Exstrophy of urinary bladder	7535	8	2	—
Atresia and stenosis of urethra	7536	17	15	—
Bilateral renal agenesis	7530	43	36	83.7
Polycystic kidneys	7531	70	64	91.4
Total major anomalies of urinary tract	—	138	117	84.8
Minor anomalies				
Unilateral renal agenesis	7530	49	39	79.6
Unilateral cyst(s) of kidneys	7531	109	100	91.7
Hydronephrosis	7532	544	508	93.4
Other kidney anomalies	7533	62	37	59.7
Other specified anomalies of ureter	7534	7	4	—
Other specified anomalies of bladder and urethra	7538	30	26	86.7
Unspecified anomalies of urinary system	7539	15	13	—
Total minor anomalies of urinary tract	—	816	727	89.1
TOTAL urinary tract anomalies	—	954	844	88.5
Musculoskeletal anomalies				
Major anomalies				
Congenital muscular dystrophy	3590	2	1	—
Myotonia	3592	2	1	—
Arthrogryposis	7558	21	9	42.9
Chondrodysplasia	7564	43	39	90.7
Osteodystrophy	7565	29	26	89.7
Anomalies of diaphragm (hernia)	7566	88	51	58.0
Congenital defect of abdominal wall	7567	158	129	81.6
Major skin anomalies	7571; 7573	6	1	—
Total major musculoskeletal anomalies	—	349	257	73.6
Minor anomalies				
Spine anomalies	7542	6	3	—
Deformities of feet	7545-7547	279	48	17.2
Thorax and upper limb deformities	7548	9	4	—
Polydactyly	7550	84	8	9.5
Syndactyly	7551	69	2	2.9
Reduction deformities of upper limbs	7552	79	18	22.8
Reduction deformities of lower limbs	7553	31	11	35.5
Other anomalies of upper limbs	7555	17	3	—
Other anomalies of lower limbs	7556	12	1	—
Other unspecified anomalies of limbs	7559	24	9	37.5
Skull and face anomalies	7560	48	10	20.8
Spine anomalies	7561	22	6	27.3
Anomalies of ribs and sternum	7563	6	0	—
Other musculoskeletal anomalies	7568; 7569	5	2	—
Total minor musculoskeletal anomalies	—	694	125	18.0
TOTAL musculoskeletal anomalies	—	1043	382	36.6
Miscellaneous				
Cleft lips and palates				
Cleft palate	7490	72	1	1.4
Cleft lip	7491	80	20	25.0

Cleft palate with cleft lip	7492	164	36	22.0
Total cleft lips and palates	—	316	57	18.0
<b>Respiratory system anomalies</b>				
<b>Major anomalies</b>				
Anomalies of larynx, trachea, and bronchus	7483	3	0	—
Congenital cystic lung	7484	11	11	—
Total major respiratory system anomalies	—	14	11	—
<b>Minor anomalies</b>				
Anomalies of nose	7481	4	1	—
Hypoplasia and dysplasia of lung	7485	3	1	—
Other anomalies of lung	7486; 7488	9	8	—
Total minor respiratory system anomalies	—	16	10	—
TOTAL respiratory system anomalies	—	30	21	70.0
<b>Other anomalies of head and neck</b>				
Micrognathia	5240	14	1	—
Retrognathia	5241	5	0	—
Webbing of neck	7445	3	0	—
Other anomalies of face and neck	7448; 7449	20	5	25.0
TOTAL other anomalies of head and neck	—	42	6	14.3
<b>Congenital anomalies of ear and eye</b>				
Anophthalmos	7430	5	0	—
Microphthalmos	7431	7	2	—
Anomalies of eyelids, lacrimal system, and orbit	7436	3	1	—
Other specified anomalies of eye	7438	1	0	—
Unspecified anomalies of eye	7439	2	1	—
Anomalies of ear causing impairment of hearing	7440	3	0	—
Other specified anomalies of ear	7442	3	0	—
Unspecified anomalies of ear	7443	2	0	—
TOTAL congenital anomalies of ear and eye	—	26	4	15.4
Hemangioma any site	2280	7	1	—
Lymphangioma (neck)	2281	101	98	97.0
<b>Miscellaneous benign neoplasms</b>				
Dyshormogenetic goiter	2461	2	2	—
Coccyx teratoma	6537	14	11	—
TOTAL miscellaneous benign neoplasms	—	126	112	88.9
Ovarian cyst	7520	38	38	100.0
TOTAL all anomalies	—	4615	2593	56.2

The best detected abnormalities were those of the urinary system (88.5%) and central nervous system (88.3%). Cardiac abnormalities were not well detected, whether major (38.8%) or minor (20.8%). The lowest rates of detection were for the minor abnormalities of the musculoskeletal system (18% vs 73.6% for the major ones) and for cleft lips and palates (18.0%).

#### Gestational age on diagnosis.

For all cases a diagnosis was made at  $25.8 \pm 7.5$  weeks ( $24.2 \pm 7.2$  weeks for the major abnormalities and  $27.6 \pm 7.4$  weeks for the minor ones). The fetuses bearing >1 malformation received diagnoses earlier than those with a single malformation (23.1 weeks vs 25.9 weeks;  $P < .001$ ). The gestational age at diagnosis depended on the type of malformation (Table II).

**Table II.** Average gestational age at diagnosis for isolated malformations

Anomaly	No.	Gestational age (wk)	SD	Before 24 wk* (%)
Central nervous system anomalies				
Anencephaly (7400)	128	19.1	4.5	82.8
Encephalocele (7420)	21	22.8	7.5	66.7
Hydrocephalus (7423)	110	27.9	7.2	35.5
Other central nervous system anomalies	144	25.7	7.2	42.3
Heart and great vessel anomalies				
Major anomalies	76	26.8	7	40.7
Minor anomalies	48	30.7	5.5	5.1
Digestive system anomalies	85	30.2	5.8	12.5
Urinary tract anomalies				
Major anomalies	66	23	6.5	56.8
Minor anomalies	582	29.4	6.5	27.7
Musculoskeletal anomalies				
Major anomalies	173	23.4	7.2	62.9
Minor anomalies	48	23.4	5.3	59.6
Miscellaneous				
Cleft palates and cleft lips	40	28	7.4	31.6
Hemangioma any site (2281)	78	17	4.8	92.3
Ovarian cyst (7520)	37	34.2	3.2	0
TOTAL isolated anomalies	1675	26.4	7.4	41.1

Details concern malformations for which average age at diagnosis differs from whole category.

\*Rate of diagnosis before 24 weeks.

Diagnosis was made at an earlier time for the minor and major musculoskeletal abnormalities ( $23.4 \pm 6.5$  weeks), the abnormalities of the central nervous system ( $24.1 \pm 7.4$  weeks), and for the major urinary abnormalities ( $23.0 \pm 6.5$  weeks). The cardiac abnormalities (major and minor) were detected later in pregnancy ( $28.1 \pm 6.8$  weeks), as were the cleft lips and palates ( $28.0 \pm 7.4$  weeks), the minor urinary abnormalities ( $29.4 \pm 6.8$  weeks), and those abnormalities of the digestive system ( $30.2 \pm 5.8$  weeks).

The anencephalies and the cystic hygromas of the neck were detected at the earliest times.

Overall, 44% of the malformed fetuses with abnormalities (isolated or associated) were detected before 24 weeks. For the fetuses with severe malformations this proportion rose to 55%.

#### Outcome of malformed fetuses.

Among the cases detected during pregnancy, 12% led to spontaneous abortions or death in utero, and 27% led to an elective termination. For the severe malformations there were 15% spontaneous abortions and deaths in utero and 41% elective terminations. Eighty-three percent of the terminations were carried out before 24 weeks.

Only 27% of the infants with a severe malformation detected before birth were born alive versus 76% when the diagnosis was made after birth. We also noted a difference, albeit smaller, for the minor malformations (85% vs 95% live births for prenatal and postnatal diagnoses, respectively).

#### False positives and false alarms.

All the diagnoses or suspected cases of malformations during pregnancy in the participating centers were recorded on initial detection. Some diagnoses were made in fetuses who were in fact found to be normal. In some cases the diagnostic error was rectified on subsequent ultrasonographic examination at a later stage of the pregnancy. These cases were referred to as false alarms. The false positives were the cases in which the error was revealed only at term.

Of the 3085 diagnoses of malformation made during pregnancy, 2593 (84%) were true positives, 187 (6%) were false alarms, and 305 (9.9%) were false positives. The details of the false positives and false alarms are listed in Table III.

**Table III.** False positives and false alarms

	ICD-9	False positive		False alarm		True positive (No.)
		No.	%	No.	%	
<b>Central nervous system anomalies</b>						
Spina bifida without hydrocephalus	7419	2	3.1	4	6.2	59
Reduction deformities of brain	7422	1	2.6	1	2.6	37
Microcephalus	7421	7	19.4	3	8.3	26
Hydrocephalus	7423	21	9.1	21	9.1	188
Other anomalies of brain	7424	9	9.5	27	28.4	59
<b>TOTAL central nervous system anomalies</b>	<b>7424; 7429</b>	<b>40</b>	<b>5.3</b>	<b>56</b>	<b>7.5</b>	<b>652</b>
<b>Heart and great vessel anomalies</b>						
Transposition of great vessels	7451	0	0.0	2	12.5	14
Hypoplastic left heart syndrome	7467	1	4.0	0	0.0	24
Other anomalies of aorta	7471; 7472	4	80.0	0	0.0	1
Ventricular septal defect	7454	6	11.8	3	5.9	42
Atrial septal defect	7455	2	13.3	0	0.0	13
Other specified anomalies of heart	7468	7	12.7	4	7.3	44
Unspecified anomalies of heart	7469	8	22.9	9	25.7	18
<b>TOTAL heart and great vessel anomalies</b>	<b>—</b>	<b>28</b>	<b>9.0</b>	<b>18</b>	<b>5.8</b>	<b>264</b>
<b>Digestive anomalies</b>						
Esophageal atresia and stenosis	7503	4	12.1	0	0.0	29
Atresia of small intestine	7511	3	4.7	1	1.6	60
Occlusion	7512	1	9.1	2	18.2	8
All other digestive anomalies	—	16	32.0	8	16.0	26
<b>TOTAL digestive anomalies</b>	<b>—</b>	<b>24</b>	<b>15.2</b>	<b>11</b>	<b>7.0</b>	<b>123</b>
<b>Urinary system anomalies</b>						
Bilateral renal agenesis	7530	2	5.1	1	2.6	36
Polycystic kidneys	7531	3	4.4	1	1.5	64
Unilateral renal agenesis	7530	2	4.8	1	2.4	39
Unilateral cyst(s) of kidneys	7531	9	8.0	3	2.7	100
Hydronephrosis	7532	146	21.0	42	6.0	508
Other kidney anomalies	7533	4	9.5	1	2.4	37
Other anomalies of bladder and urethra	7536; 7538	6	17.1	3	8.6	26
Unspecified anomalies of urinary system	7539	5	25.0	2	10.0	13
<b>TOTAL urinary system anomalies</b>	<b>—</b>	<b>177</b>	<b>16.5</b>	<b>54</b>	<b>5.0</b>	<b>844</b>
<b>Musculoskeletal anomalies</b>						
Chondrodysplasia	7564	3	6.8	2	4.5	39
Anomalies of diaphragm (hernia)	7566	3	5.4	2	3.6	51
Laparoschisis	7567	0	0.0	5	3.7	129
Deformities of feet	7545-7547	9	15.5	1	1.7	48
Anomalies of limbs	7550-7556	4	6.7	4	6.7	52
Skull and face anomalies	7560	1	8.3	1	8.3	10
Other anomalies of face and neck	7448; 7449; 7561	1	4.8	9	42.9	11
<b>TOTAL musculoskeletal anomalies</b>	<b>—</b>	<b>21</b>	<b>4.9</b>	<b>24</b>	<b>5.6</b>	<b>382</b>
<b>Respiratory system anomalies</b>						
Cleft lips and palates	7490; 7492	0	0.0	0	0.0	57
Hemangioma any site	2281	7	5.8	15	12.5	98
Miscellaneous	—	7	—	8	—	—
<b>TOTAL anomalies</b>	<b>—</b>	<b>305</b>	<b>9.9</b>	<b>187</b>	<b>6.1</b>	<b>2593</b>

Rates are expressed for the total number of diagnoses of malformations. Details are given for all cases of false positives and false alarms.

Of these 305 false-positive malformations incorrectly diagnosed, 49 were in a fetus bearing another correctly identified malformation, and in 256 cases this was the only abnormality incorrectly diagnosed in a normal fetus. Among these misdiagnoses only 1 had a termination of pregnancy. It was a severe growth restriction with oligoamnios, the fetus weighing 200 g at 26 weeks.

Comment

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The Eurofetus Study included 3685 fetuses with structural malformations and represents the largest study to date on the evaluation of ultrasonographic screening for such defects. In view of the diversity of congenital malformations and the differences in performance of ultrasonographic diagnosis as a function of the type of malformation, only a survey of a large population can provide reliable data on routine ultrasonographic screening.

In our study the overall sensitivity of prenatal ultrasonography for correctly identifying fetuses with structural defects was 61.4%. Other reports in the literature for screening malformations in the general population<sup>1-9</sup> showed a range from 8.7% to 78.3%. However, differences in the criteria used for recording malformations make it difficult to compare the different studies. Some authors have excluded a priori abnormalities that are considered undetectable by ultrasonography, which may differ among centers. Second, in small series there may be chance differences in the rate of the various types of malformation that are not detected with equal ease. These differences will lead to differences in the level of detection of malformations. The series reported by Luck,<sup>9</sup> whose sensitivity was as high as 85% before 19 weeks' gestation, was considered to be unrepresentative<sup>10</sup> because of the abnormally high proportion of hydronephroses, which are among the best detected.

Furthermore, the best sensitivities can be obtained in centers using the most recent equipment and on populations at high risk. For instance, >10 years ago Sabbagha et al<sup>11</sup> reported a sensitivity of 95% for ultrasonographic targeted imaging for fetal abnormalities in women at high risk for birth defects. This excellent sensitivity was almost certainly biased by the exclusion of cardiac abnormalities. Another factor may also influence the overall level of detection of malformations, namely, the number of ultrasonographic examinations carried out during pregnancy. In our study we estimated a mean number of ultrasonographic examinations per pregnancy of 3 for the fetuses with undiagnosed malformations, which is regarded as the norm for ultrasonographic screening in Europe. Some authors recommend a single ultrasonographic examination performed in the second trimester. However, this may prevent detection of certain abnormalities that become apparent later in the pregnancy and may thus reduce the overall reliability of ultrasonographic screening. In our series 38.5% of the malformations were detected after 29 weeks. This late detection involved mostly minor abnormalities (53.8% of minor abnormalities detected after 29 weeks vs 24.8% for the major ones).

In contrast to predictive value, the sensitivity should be similar in any given population and be unaffected by the prevalence of the disease. With respect to ultrasonographic screening for malformations, the awareness of certain risk factors may well motivate more detailed examination of the fetus, which would thereby lead to an increase in efficacy. This, however, should not be used as an argument to restrict ultrasonographic screening to populations at risk, because most congenital malformations occur in patients with no known risk factors,<sup>7</sup> and clinical signs may be absent or arise late in pregnancy.

In a large series major and minor cardiac abnormalities, cleft palates, malformations of the foot, limb shortening, and severe ocular defects such as anophthalmia (none of 5 cases was detected in our population) were not quickly identified. However, these malformations can be detected by ultrasonography in the right circumstances, and perhaps some of these cases could have been identified by better trained echographers (especially for abnormalities of the heart and large vessels) or by more systematic examination (for instance, measurement of limb length, search for image of lens or lips on examination of fetal face).

We included in our survey only those abnormalities that were detectable at birth or on abortion (elective or spontaneous). With respect to the abortions and deaths in utero, the presence of a malformation was verified on autopsy in 90% of cases. For the malformations noted at birth, we did not record those detected after the postpartum hospitalization period. However, it is known that many congenital malformations, especially those of the heart and digestive tract, become apparent in the first months of life or even later. From the data in the Eurocat register, approximately 10% of malformations are detected after the first postnatal week. Therefore our overall screening sensitivity may be overestimated slightly. However, it should be borne in mind that the malformations appearing later in life are generally of a minor nature.

With respect to the consequences of antenatal screening on obstetric management, 2 elements must be taken into consideration: the severity of the malformations detected and the gestational age. Like other authors, we found that the major defects are detected more quickly than the minor ones. Overall, 3 of 4 of the major malformations were detected by ultrasonographic examination. Our results, which are close to those from other studies,<sup>3,12</sup> are relevant to subsequent management. In our study the incidence of live births was significantly lower for the cases where defects were detected during pregnancy, especially for the major malformations. This difference is caused by the fact that many of these pregnancies were terminated after the defect was detected. These findings are in agreement with those of the randomized study of Saari-Kemppainen et al.<sup>2</sup>

It is important to consider the age at which terminations are carried out, because there are considerable differences between countries. In countries such as France, when a defect is considered to be incurable and particularly severe, termination at the request of the parents is authorized by law at any stage of pregnancy. In many other countries, including the United States, terminations are illegal after viability. In our study the diagnosis of malformation was made before 24 weeks in only 44% of cases for all malformations and in 55% of cases for the severe defects. In our population 83% of elective terminations were carried out before 24 weeks. Although 1 author<sup>3</sup> reported higher rates for early diagnosis than ours, most studies have found similar<sup>7</sup> or lower<sup>1,6,12</sup> rates. These late diagnoses generally are not caused by delays in performing ultrasonographic examinations. In our population 93% of patients benefited from at least 1 ultrasonographic examination before 24 weeks, and there was no difference in frequency of examinations before 24 weeks between the undetected and detected cases. The malformations detected after 24 weeks were either those with late expression such as those of the digestive system or those overlooked on first examination and picked up on subsequent ones.

In conclusion, in this survey of a large number of malformations, we were able to determine with reasonable accuracy the reliability of antenatal ultrasonographic screening of malformations in an unselected population. The results are interesting from 3 points of view. Knowledge of the performance of screening of major abnormalities and the gestational age at diagnosis is particularly valuable for decisions on management during pregnancy, especially for the severe cases for which the parents may request termination. Data on the limits of diagnosis can also provide objective information to parents who inquire about the ability to detect fetal abnormalities in their offspring. Last, areas in which echographers should make efforts to improve the quality of screening for fetal malformations are highlighted.

The following persons and institutions constituted the Eurofetus Study Group.

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