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Screening for foetal malformations: performance of routine ultrasonography in the population of the Swiss Canton of Vaud

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Summary

Objective: To determine the sensitivity of ultrasonography in screening for foetal malformations in the pregnant women of the Swiss Canton of Vaud.

Study design: Retrospective study over a period of five years.

Method: We focused our study on 512 major or minor clinically relevant malformations detectable by ultrasonography. We analysed the global sensitivity of the screening and compared the performance of the tertiary centre with that of practitioners working in private practice or regional hospitals.

Results: Among the 512 malformations, 181 (35%) involved the renal and urinary tract system, 137 (27%) the heart, 71 (14%) the central nervous system, 50 (10%) the digestive system, 42 (8%) the face and 31 (6%) the limbs. Global sensitivity was 54.5%. The lowest detection rate was observed for cardiac anomalies, with only 23% correct diagnoses. The tertiary centre achieved a 75% detection rate in its outpatient clinic and 83% in referred patients. Outside the referral centre, the diagnostic rate attained 47%.

Conclusions: Routine foetal examination by ultrasonography in a low-risk population can detect foetal structural abnormalities. Apart from the diagnosis of cardiac abnormalities, the results in the Canton of Vaud are satisfactory and justify routine screening for malformations in a low-risk population. A prerequisite is continuing improvement in the skills of ultrasonographers through medical education.

Key words: congenital malformations; ultrasound screening; prenatal diagnosis

Introduction

Like many medical technologies, prenatal ultrasonography was introduced as a routine procedure without scientific evidence that it improves foetal and neonatal morbidity and mortality. Ultrasound scans are now an integral part of routine follow-up of pregnancy in most European countries.

Since the eighties, ultrasonographic equipment has improved to such a degree that the sensitivity of ultrasonographic screening for malformations in the latest European studies is around 60%, while specificity is as high as 99%. Despite a low prevalence (2–3%), structural malformations account for 25% of perinatal deaths.

Ultrasound scans may provide important information about foetal anatomy. Parents should be aware of the goals and limitations of this type of examination. In the event of lethal or severe malformations, they will face a choice between proceeding with the pregnancy or considering termination. Ultrasound examination may make it possible to plan birth in a tertiary centre ensuring adequate neonatal management. In some cases prenatal ultrasound may result in an unnecessary burden of anxiety, but it should be stressed that in the vast majority of cases its reassuring effect is of very significant benefit to the patient.

The pertinence of screening for congenital malformations is debated. Randomised studies report conflicting results in evaluating morbidity and mortality in a screened population [1, 2]. Moreover, the rate of detection varies widely, depending among other things on the organs considered (6% to 90%) [3, 4]. Results reported by a referral centre dealing with high-risk patients cannot be compared with those in the general population.

In Switzerland, payment for these examinations by health insurance schemes has been challenged. The purpose of this study is to describe the performance of ultrasound screening in the detection of major foetal malformations during a five-year period in a general population.

Patients and methods

In the Swiss Canton of Vaud, 38110 births were registered between January 1994 and December 1998. During this period, pregnant women generally underwent an early scan at 11–14 weeks' gestation, a second at 20–22 weeks and a third at 32–34 weeks. Scans were performed by obstetricians, radiologists and general practitioners either in private practice or hospitals. The only tertiary centre in the Canton of Vaud deals with a high-risk population but also screens low-risk patients from its prenatal clinic.

Table 1
Detection of foetal abnormalities by organs involved.

Abnormalities	True positive	False negative
Central nervous system	*58*	*13*
Hydrocephalus	25	8
Anencephaly	16	1
Spina bifida	15	4
Encephalocele	2	0
Heart abnormalities	*31*	*106*
Atrioventricular canal defect	3	9
Hypoplastic left and right heart syndrome	7	2
Univentricular heart	1	3
Transposition of great vessels	2	8
Tetralogy of Fallot	0	8
Common truncus	1	1
Double outlet ventricle	1	3
Aortic arch hypoplasia	3	6
Coarctation of aorta	1	11
Cardiomyopathy	1	8
Complex malformations	4	8
Ventricular septal defect >3 mm	6	36
Miscellaneous heart malformation	1	3
Digestive system abnormalities	*28*	*22*
Miscellanous atresia	7	10
Laparoschisis	5	1
Omphalocele	8	5
Diaphragmatic hernia	8	6
Members: reduction of limbs	*13*	*18*
Kidney and urinary tract abnormalities	*129*	*52*
Bilateral renal agenesia	4	0
Unilateral renal agenesia	9	5
Bilateral multicystic dysplasia	3	0
Unilateral multicystic dysplasia	22	1
Polycystic kidneys	2	1
Exstrophy of the bladder	1	2
Other anomalies of the kidneys	5	1
Stenosis of pyelo-ureteral jonction	33	12
Double collecting system	13	6
Pyelo-caliceal dilatation	21	8
Hydronephrosis	4	2
Anomaly of the urethra	4	4
Other anomalies of the ureter	4	9
Other anomalies of the bladder	4	1
Face	*21*	*21*
Unilateral cleft lip	15	14
Bilateral cleft lip	6	7

The Vaud registry of congenital anomalies is included in the Swiss registry of EUROCAT, a European network of population-based registries for epidemiologic surveillance of congenital anomalies. Currently more than 900,000 births per year in Europe are surveyed by 36 registries in 17 European countries. To ensure a complete and accurate database, the Vaud registry is based on multiple sources of information [5, 6]. All live births, stillbirths and terminations of pregnancy with a malformation demonstrated in the prenatal or postnatal period are included. The data are collected by one of the authors (MCA) and contain comments on prenatal ultrasonographic description of the malformations. It includes information regarding the infant, the pregnancy and the parents.

This retrospective study focuses on major and some minor malformations detectable by ultrasonography divided into 6 main groups (table 1). The correlation between ultrasound examination and postnatal or postmortem findings was considered true positive if the findings were totally or partially confirmed. Major malformations included lethal or incurable abnormalities and conditions associated with severe handicap or requiring surgery. Minor malformations were likewise considered when clinically relevant. They also afford evidence of the quality of reporting [7] and include ventricular septal defect of more than 3 mm and pyelo-caliceal dilatations.

The sensitivity of prenatal diagnosis was studied in relation to the various malformations. Since foetuses may present multiple malformations, the detection rate was also calculated in terms of malformed foetuses. In a second analysis we compared the sensitivity observed in the tertiary centre for low-risk patients with the results obtained in the rest of the Canton of Vaud. Cases referred to the tertiary centre for further examination included those referred for suspected foetal abnormality or for pregnancy follow-up in the last weeks of gestation prior to delivery at our centre. All these cases were credited to the referring practitioners and not to the centre. This evaluation was performed by matching the data of the Vaud registry with the data prospectively collected in the tertiary centre.

Fischer's exact test was used to compare detection rates in different populations. A p value <0.05 was considered significant.

Results

During the study period, 1270 foetal malformations were entered in the Vaud registry of congenital anomalies. When compared with the 38110 births, this corresponds to a prevalence of 3.3%. Of these, 512 (1.3%) were clinically relevant major and minor malformations (table 1). They were observed in 416 foetuses, some presenting with multiple malformations. The most frequently observed abnormalities involved the renal and urinary tract (181/512; 35%); among cases associated with urinary tract dilatation (stenosis of pyeloureteral junction, double collecting system, pyelocaliceal dilatation and hydronephrosis), 71/99 (72%) were detected in the prenatal period. Unilateral renal multicystic dysplasia was diagnosed in 95% of cases (22/23), and unilateral renal agenesia in 64% (9/14). Lesions resulting in terminal renal failure associated with anamnios were diagnosed in all cases (bilateral renal agenesia and bilateral multicystic dysplasia; n = 7).

Cardiac malformations accounted for 27% of all anomalies (n = 137). Major malformations noticeable in the four-chamber view were detected in 13/36 cases (36%). Among 42 cases of ventricular septal defect (>3 mm), 6 (17%) were diagnosed. The detection rate for other cardiac malformations was 12/59 (20%) and the central nervous system was involved in 71 abnormalities (14%). Most cases of neural tube defect (84%; n = 38) and hydrocephalus (76%; n = 33) were diagnosed on ultrasound examination. Other cases involved the digestive system (n = 50; 10%), the face (n = 42; 8%) and the limbs (n = 31; 6%).

Overall, foetal malformations were diagnosed during the prenatal period in 279 cases (55%) (table 2). The best detection rate was observed for central nervous system anomalies (82%), followed by renal and urinary tract (71%), digestive system (56%), face (50%), limbs (42%) and heart malformations (23%).

Considering foetuses as individuals presenting one or more malformations, the overall sensitivity of prenatal diagnosis by ultrasound was 57.2% (238/416).

Screening outside the referral centre achieved correct diagnosis in 175 of 374 cases (47%). The tertiary centre achieved a 75% detection rate among patients followed in its prenatal clinic ($p < 0.001$) and an 83% rate in referred cases (table 3). The low detection rate for heart anomalies (14% in private practice and 53% in tertiary care centres) reflects the difficulties encountered in obtaining the appropriate planes for thorough examination of the whole 3D anatomy of this organ. On the other hand, the good results (50%) obtained for a malformation reputedly difficult to see, i.e. cleft lip, show that a complete and thorough examination of a specific anomaly can improve its detection.

Table 2

Sensitivity of screening in the Canton of Vaud.

Type of malformation	sensitivity (%)
Central nervous system	82
Heart disease	23
Digestive abnormalities	56
Renal and urinary tract abnormalities	71
Facial cleft	50
Reduction of limbs	42
Global	55

Table 3

Sensitivity of screening in the tertiary centre, in private practice and in regional hospitals.

	(A)	(B)	(C)	A vs C
Type of malformation	**Tertiary centre**	**Referred * to tertiary centre**	**Non-referred and referred* to tertiary centre**	**Fischer exact test**
Central nervous system (n = 71)	85% (17/20)	94% (33/35)	80% (41/51)	0.74
Heart disease (n = 137)	58% (15/26)	48% (14/29)	14% (16/111)	<0.0001
Digestive abnormalities (n = 50)	75% (12/16)	100% (11/11)	47% (16/34)	0.07
Renal and urinary tract abnormalities (n = 181)	80% (48/60)	90% (56/62)	66% (80/121)	0.0582
Facial cleft (n = 42)	88% (7/8)	73% (11/15)	41% (14/34)	0.044
Reduction of limbs (n = 31)	56% (5/8)	100% (6/6)	35% (8/23)	0.22
Global (n = 512)	73% (104/138)	83% (131/158)	47% (175/374)	<0.0001

* Includes cases referred for suspected foetal abnormality or for pregnancy follow-up in the last weeks of pregnancy and delivery

Discussion

Prenatal ultrasonography is the best means of diagnosing malformed foetuses [8]. However, published results concerning the sensitivity of the screening vary greatly, depending on the population studied (high versus low risk), the quality of the equipment and in particular the ultrasonographers' experience. Nowadays, high-resolution equipment allows the detection of minor malformations considered to be sonographic markers of specific conditions. These were not included in our study, to maintain the homogeneity of the population screened. We focused our attention on severe abnormalities which can be detected by prenatal ultrasonography.

The detection rate for congenital abnormalities by routine ultrasonography in a general population varies from 8 to 84% [1, 2, 9–12]. Levi et al. [13] reported 40.4% sensitivity in a study performed in Belgium, Eik-Nes et al. [14] 43% in Norway, and in the recent Eurofetus study [15] the rate was 61.4%. Our detection rate of 55% is comparable to these results, as is the 47% attributed to the practitioners working outside the tertiary centre. These results are encouraging and compare very favourably with the detection rate of 22% for obstetricians in the study of Bernaschek et al. [16] and the 14% reported by Lys et al. [17]. This can probably be explained by the effort made at the beginning of the nineties in terms of post-graduate and continuous education in this field.

In our study, the data for some malformations known to be rather difficult to diagnose, such as cleft lip with or without cleft palate, show a 50% detection rate which is in agreement with the results of the Euroscan Study Group [18]. In our region, however, efficacy of screening for heart diseases is below the usual standards. Our global prenatal diagnosis rate for cardiac malformations is only 22.6% compared with 45–55% reported in the countries participating to the Euroscan Study Group [19]. Our detailed results emphasise the need for improved training in the field of heart, limb and diaphragmatic anomalies.

Awareness of our screening performance in the general population provides evidence with which to explain the limits of ultrasonographic examination to our patients and emphasises the need for a network of trained ultrasonographers to whom difficult or high-risk cases should be referred.

We believe these results justify routine ultrasonographic screening for foetal malformations in all pregnancies. To propose screening only to high-risk patients is discriminatory, for many reasons. First, it is well known that most of the malformations are discovered in the low-risk group [20]. Second, there is no universal definition of a high-risk pregnancy, and third, it is only after birth that a case can be declared low-risk with certainty. The usual high-risk patients (family history of congenital disease, previous pregnancy with abnormality) should be referred to well-trained ultrasonographers along with patients in whom ultrasonographic markers for malformations are demonstrated (oligohydramnios, polyhydramnios, foetal growth retardation, increased nuchal translucency measurement in first trimester with normal karyotype).

In conclusion, we share the view of Bucher and Schmidt [21] who in their meta-analysis insist that "routine ultrasound screening in pregnancy is indicated only if explicitly performed to exclude congenital malformations". We also agree with the conclusion of the British Columbia Office of Health Technology Assessment [22] that routine ultrasonography in a low-risk population only has sense if it forms part of a programme of prenatal diagnosis including screening for trisomy 13, 18 and 21. Finally, ultrasound screening should be linked to an improved detection rate for foetal malformations through appropriate continuous education, and should always be associated with the use of informed consent protocols.

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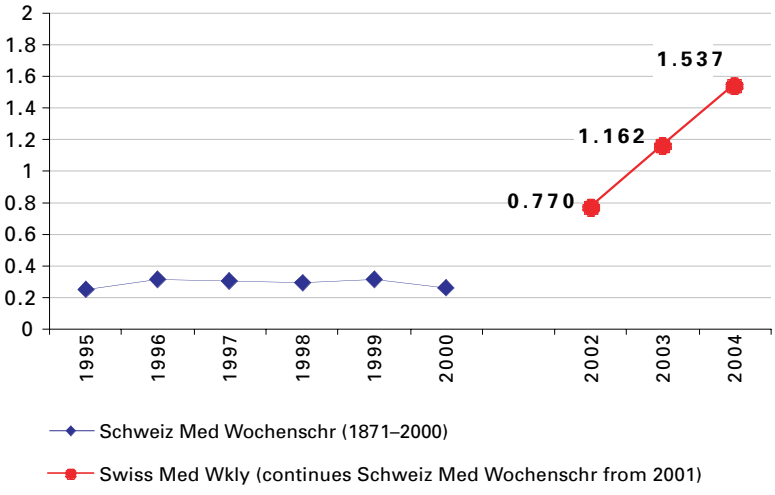

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