Original Article

VALIDATION OF ULTRASOUND DIAGNOSIS OF FETAL ANOMALIES AT A SPECIALIST CENTER

CARLOS NORONHA NETO[1]*, ALEX SANDRO ROLLAND DE SOUZA[2], OLÍMPIO BARBOSA DE MORAES FILHO[3], ADRIANA MOTA BIONE NORONHA[4]
Study conducted at Instituto de Medicina Integral Professor Fernando Figueira – I.M.I.P., Recife, PE, Brazil

ABSTRACT

Objective. To validate ultrasound diagnoses of fetal anomalies made at a specialist Fetal Medicine Center in Pernambuco, Brazil.

Methods. A cross-sectional study was performed to validate diagnostic test results, including all high-risk pregnant women submitted to morphological obstetric ultrasound at the *Instituto de Medicina Integral Professor Fernando Figueira* (I.M.I.P.), from March 2002 to March 2006. Prenatal diagnoses were confirmed after birth. Variables analyzed were sociodemographic characteristics and prenatal and postnatal frequencies of fetal anomalies. Agreement between prenatal and postnatal diagnoses of congenital anomalies was evaluated using the Kappa index. Youden's index was used to validate prenatal ultrasound diagnoses.

Results. Nine hundred and eighty-nine patients were eligible for the study and of these 457 expectant mothers were included. Mean maternal age was 24.8 ± 6.5 years. Prenatal ultrasonography diagnosed congenital anomalies in 289 (63.2%) patients, 257 (56.2%) of which were confirmed after birth. Comparing the prenatal diagnoses of congenital anomalies with the postnatal results, prenatal ultrasound diagnosis of fetal anomalies exhibited 96% sensitivity and 79% specificity, good agreement (K=0.76) and good diagnostic validity (Y=0.75).

Conclusions. Prenatal diagnoses of fetal anomalies made by morphological ultrasonography at a specialist Fetal Medicine center in Pernambuco exhibited good sensitivity, specificity, prenatal to postnatal concordance, and diagnostic validity.

KEYWORDS: Ultrasonography. Sensitivity and specificity. Congenital, hereditary and neonatal diseases and anomalies. Prenatal diagnosis.

***Correspondence:**
Rua da Angustura, nº 195 –
Apto. 204 - Aflitos
Recife – PE
52050-340
Telefone: (81) 32415876
ca.no.ne@hotmail.com

INTRODUCTION

The progress that has been made in the field of ultrasonography has contributed to an increase in the detection of fetuses with structural anomalies both among low-risk[1] and high-risk populations.[2-4] The great potential of ultrasonography for screening for morphological abnormalities throughout all trimesters of the pregnancy[1] has meant that its use with obstetric patients is becoming a routine part of prenatal care.[5]

Recent hospital-based research, covering a short time period, reported a 2.6% prevalence of congenital anomalies among the study population.[4] Although the accuracy of ultrasound for the diagnosis of congenital malformations has been the subject of many studies, it has been fo und that low sensitivity in combination with low rates of false-positives was associated with tracking low-risk pregnancies, leading to the belief that ultrasonography is most applicable to pregnancies involving fetal abnormalities and/or high levels of risk.[4]

The majority of studies located were carried out with patients in hospital and reported high rates of detection and an elevated incidence of major malformations.[6-8] However, a population study carried out over a long period found a low level of sensitivity (28.4%), although detection of certain structural anomalies was relatively good.[4]

The morphological ultrasound scan, performed in the second trimester of the pregnancy, and the continuing specialization of ultrasonographists, have increased the likelihood that congenital malformations will be detected, increasing diagnostic sensitivity.[9] In certain studies, the sensitivity of detection of fetal anomalies, before the 24th week of gestation, was 93% for the central nervous system, 45.2% for the circulatory system, 85.2% for the digestive system, 85.7% for the urinary system, 84.6% for the musculoskeletal system and 95.2% for other anomalies found. Therefore, it is suggested that ultrasonography between the 20th and 22nd weeks of pregnancy can detect the majority of congenital anomalies.[10]

1. Mestrado em Tocoginecologia e Médico do Instituto de Medicina Integral Professor Fernando Figueira – IMIP, Recife, PE
2. Mestre em Saúde Materno Infantil e Médico pelo do Instituto de Medicina Integral Professor Fernando Figueira – IMIP, Recife, PE
3. Doutor em Tocoginecologia e médico da Maternidade Monteiro de Moraes do Centro Integrado à Saúde Amaury de Medeiros – UPE, Recife, PE
4. Especialização em UTI obstétrica e Enfermeira do Instituto de Medicina Integral Professor Fernando Figueira – IMIP, Recife, PE

The RADIUS and Eurofetus studies found evidence that, when compared with basic healthcare centers, centers specialized in fetal medicine had a better diagnostic approach to fetal anomalies before the 24th week of gestation. Notwithstanding, collaborative studies are needed to establish the true levels of sensitivity and specificity achieved by ultrasound diagnosis at a large number of hospitals.[11]

In this context, it is suggested that validation of the prenatal diagnosis of congenital anomalies is dependent on the institution studied, the equipment used and, primarily, on the ultrasonographist. Therefore, it was necessary to undertake a study that would determine the validity of prenatal ultrasound diagnosis of fetal anomalies as performed at a specialist fetal medicine center in the State of Pernambuco, Brazil.

Methods

This was a cross-sectional observation study to validate a diagnostic test carried out with pregnant women in the high-risk ward at the *Instituto de Medicina Integral Professor Fernando Figueira* (I.M.I.P.) between March 2002 and March 2006.

The sample size calculation was performed using the STATCALC function in Epi-Info 2007, version 3.4.1, with a predicted frequency of congenital malformations, among high-risk gestations diagnosed during the prenatal period, of 27%[11] and a relative accuracy of 20%. This resulted in a sample of 445 expectant mothers for a confidence level of 99%.

The study included all expectant mothers who underwent at least one morphological ultrasound scan at the Fetal Medicine department at the IMIP at a gestational age greater than or equal to 22 weeks and/or birth weight greater than or equal to 500g. Multiple births, births not taking place at IMIP and cases where the infants' medical records were missing were excluded.

F fetal morphological ultrasonography was carried out using a Toshiba SSA-350A (Corevision) ultrasound machine and a 5MHz sector transducer. Patients were examined in dorsal decubitus, with the bladder empty.

During the period studied, 989 patients were identified as candidates for inclusion on the basis of having undergone fetal morphological ultrasonography. From this number, 457 patients were recruited after application of the inclusion and exclusion criteria. Using the hospital records, all of the mothers were followed up to birth, and the newborn infants until confirmation or not of the intrauterine diagnosis of congenital anomaly. The congenital anomalies were defined according to the 10th revision of the International Classification of Diseases.[12]

Data were collected by the researcher, using the patient records from the Fetal Medicine department, in addition to the records from obstetrics and pediatrics. The newborn infants' medical records were used to investigate their postnatal diagnoses. Internal anomalies diagnosed during the prenatal period were confirmed on the basis of supplementary test results and/or clinical and surgical assessment. External anomalies were confirmed on the basis of the clinical examination performed by the neonatologist. The diagnoses for all of the patients studied were confirmed or ratified retrospectively.

Data were analyzed using Epi-Info 2007, version 3.4.1. and OpenEpi, version 2.2. The Kappa index (K)[13] was used to demonstrate whether there was concordance between the prenatal ultrasound diagnosis and postnatal result. The prenatal ultrasound diagnosis was validated by applying the Youden test (Y)[14]. The Pan American Health Organization in conjunction with the National Health Foundation (*Fundação Nacional de Saúde*)[15]. has constructed a concordance scale for these indicators. Scores of 0.00 indicate absence of concordance; from 0.01 to 0.20, concordance is weak; from 0.21 to 0.40, concordance is acceptable; from 0.41 to 0.60, concordance is regular; from 0.61 to 0.80, concordance is good; from 0.81 to 0.99, concordance is excellent and 1.00 indicates perfect concordance. The prenatal and postnatal frequencies of fetal abnormalities having thus been established, broken down by organ and system, the sensitivity and specificity of the intrauterine ultrasound diagnoses were then calculated.

The research protocol was approved by the Research Ethics Committee at the IMIP and by the National Research Ethics Commission (*Comissão National de Ética em Pesquisa,* CONEP, protocol number 901/2006. Brasília, 13 September, 2006).

Results

During the period studied, 457 high-risk expectant mothers were recruited. Prenatal ultrasonography led to a diagnosis of congenital anomaly in 289 (63.2%) patients and 257 (56.2%) of these diagnoses were confirmed postnatally.

The mothers' ages varied from 13 to 47 years, with a mean of 24.8±6.5 years. Two hundred and fifty-seven (56.2%) expectant mothers said they had no form of employment whatsoever, while 203 (44.4%) stated that their family income was between one and three times the national minimum wage. The mean gestational age at birth was 35.9±3.7 weeks.

Morphological examinations were carried out according to routine practice at the institute between the 22nd and 24th weeks, between the 26th and 28th weeks and between the 32nd and 34th weeks of the pregnancy, with the number of times each patient was examined varying from one to three. The majority of the expectant mothers underwent their first ultrasound examination between the 26th and 28th weeks.

It was observed that 289 fetuses had had a prenatal anatomic diagnosis and that of these 247 were confirmed by postnatal examination. Therefore the fetal abnormality was confirmed in 85.5% of cases with abnormal ultrasound findings. Among the cases with normal ultrasound scans, 94% of the infants did not exhibit abnormalities after birth. According to the Kappa[13] and Youden[14] indexes, the ultrasound diagnoses of congenital anomalies had good concordance with the postnatal results (K=0.76) and good diagnostic validity (Y=0.75). Sensitivity was 96% and specificity was 79% (Table 1).

Breaking down the prenatal diagnoses of congenital anomalies by body system, 129 (92.8%) of the 139 central nervous system abnormalities diagnosed postnatally had been diagnosed in advance by intrauterine ultrasound. Postnatal assessments of complications of placenta, cord, and membranes confirmed abnormalities found by ultrasound in 127 (90%) cases. Abnormalities of the genital and urinary (n=70), musculoskeletal (n=46), digestive (n=45) and circulatory (n=42) systems were confirmed postnatally with frequencies varying from approximately 73% to 87%.

Table 1. Validation of prenatal anatomic diagnoses at the Instituto de Medicina Integral Professor Fernando Figueira

Prenatal diagnosis	Congenital anomaly				Percentage (%)				
	Present		Absent						
	N	%	N	%	S	E	Y	K	p
Anatomic: Present	247	85.4	42	14.6	96	79	0.75	0.76	0.046
Absent	10	6.0	158	94.0					
Total	257	100.0	200	100.0					

S = Sensitivity, E = Specificity, Y = Youden index, K = Kappa index

Table 2. Validation of prenatal diagnoses of congenital anomalies made at the Fetal Medicine department of the Instituto de Medicina Integral Professor Fernando Figueira, broken down by major systems

Prenatal diagnosis		Congenital anomaly				Percentage (%)				
		Present		Absent						
		N	%	N	%	S	E	Y	K	p
Nervous system:	Present	129	92.8	10	7.2	99	96	0.95	0.94	0.047
	Absent	1	0.4	317	99.6					
Placenta, cord, and membranes:	Present	127	90.0	14	10.0	96	98	0.94	0.88	0.047
	Absent	2	0.7	314	99.3					
Genital and urinary systems:	Present	61	87.1	9	12.9	95	97	0.92	0.89	0.047
	Absent	3	0.8	384	99.2					
Musculoskeletal system:	Present	40	86.9	6	13.1	85	98	0.83	0.84	0.047
	Absent	7	1.8	404	98.2					
Digestive system:	Present	33	73.3	12	26.7	100	97	0.97	0.83	0.046
	Absent	0	0.0	412	100.0					
Circulatory system:	Present	31	73.8	11	26.2	100	97	0.97	0.84	0.046
	Absentt	0	0.0	415	100.0					
Abdominal wall:	Present	21	100.0	0	0.0	95	100	0.95	0.97	0.047
	Absent	1	0.3	435	99.7					
Face:	Present	13	100.0	0	0.0	76	100	0.76	0.86	0.046
	Absentt	4	1.0	440	99.0					
Soft tissues:	Present	13	86.6	2	13.4	92	99	0.91	0.89	0.047
	Absent	1	0.3	441	99.7					
Tumors:	Present	3	50.0	3	50.0	100	98	0.98	0.66	0.044
	Absent	0	0.0	451	100.0					

S: Sensitivity; E: Specificity; Y = Youden index, K = Kappa index

When the concordance and validity of the prenatal ultrasound examinations were calculated according to the infants' definitive diagnoses, it was observed that concordance and validity were excellent for placenta, cord, and membranes (K=0.88 and Y=0.94), defects of the abdominal wall (K=0.97 and Y=0.95), soft tissues (K=0.89 and Y=0.91), the circulatory (K=0.84 and Y=0.97), digestive (K=0.83 and Y=0.97), genital and urinary (K=0.89 and Y=0.92), musculoskeletal (K=0.84 and Y=0.83) and central nervous systems (K=0.94 and Y=0.95) (Table 1). Prenatal diagnosis of tumors exhibited good concordance according to the Kappa index (K=0.66) and excellent validity according to the Youden index (Y=0.98), while for facial anomalies, concordance was excellent according to the Kappa index (K=0.86) and validity was good according to the Youden index (Y=0.76) (Table 2).

The sensitivity of ultrasonography was 100% for anomalies of the digestive and circulatory systems, 99% for anomalies of the central nervous system, 96% for placenta, cord, and membranes, 95% for the genital and urinary systems and also for defects of the abdominal wall, 92% for soft tissues, 85% for the musculoskeletal system and 76% for facial anomalies. Specificity was 100% for defects of the abdominal wall and facial anomalies, 99% for anomalies of soft tissues, 98% for placenta, cord, and membranes and the musculoskeletal system, 97% for the genital and urinary, digestive and circulatory systems and 96% for the central nervous system (Table 2).

The most common congenital abnormities found postnatally were ventricular dilatation (n=90) followed by neural tube defects (n=50) and hydronephrosis (n=35). Postnatal evidence was found of 91.6% of the cases of anomalies of the lower limbs, such as clubfoot, that had previously been diagnosed by ultrasonography. All cases of congenital diaphragmatic hernia, ascites, gastroschisis, hydrops fetalis and cleft lips/palates were confirmed after birth. The anomaly that was least often confirmed postnatally was esophageal atresia (58.3%) (Table 3).

Good concordance between ultrasound and postnatal results was found for esophageal atresia (K=0.73) and good validity for diagnoses of cleft lips/palates (Y=0.75) and excellent-to-perfect concordance for all other anomalies, according to the Kappa index (Table 3).

The most common congenital anomalies of each system were analyzed. The prenatal ultrasound diagnosis had a sensitivity of 100% for single umbilical artery, renal agenesis, esophageal atresia, cardiomegaly, ascites and hydrops fetalis; of 97% for ventricular dilatation/hydrocephalus; of 96% for open neural tube defects; of 94% for hydronephrosis; of 93% for diaphragmatic hernia; of 90% for omphalocele; of 88% for complex heart disease; of 81% for clubfoot and gastroschisis; and of 75% for cleft lip/palate. Specificity was 100% for diaphragmatic hernia, gastroschisis, cleft lips/palates, ascites and hydrops fetalis; 99% for open neural tube defects, single umbilical artery, hydronephrosis, renal agenesis, clubfoot, cardiomegaly, complex heart disease and omphalocele; and 98% for ventricular dilatation/hydrocephalus and esophageal atresia (Table 3).

Table 3. Validation of prenatal diagnoses of congenital anomalies made at the Fetal Medicine department of the Instituto de Medicina Integral Professor Fernando Figueira, broken down by anatomic abnormality diagnosed anomalies

Prenatal diagnosis	Congenital anomaly				Percentage (%)				
	Present		Absent						
	N	%	N	%	S	E	Y	K	p
Ventricular Dilatation: Present	88	92.6	7	7.4	97	98	0.95	0.93	0.047
Absent	2	2.3	360	98.0					
ONTD: Present	48	96.0	2	4.0	96	99	0.95	0.95	0.047
Absent	2	0.5	405	99.5					
Hydronephrosis: Present	33	91.6	3	8.4	94	99	0.93	0.92	0.047
Absent	2	0.5	419	99.5					
Clubfoot: Present	22	91.6	2	8.4	81	99	0.90	0.85	0.047
Absent	5	1.2	428	98.8					
Diaphragmatic hernia Present	14	100.0	0	0.0	93	100	0.93	0.96	0.047
Absent	1	0.3	442	99.7					
Ascites: Present	12	100.0	0	0.0	100	100	1.00	1.00	0.047
Absent	0	0.0	445	100.0					
Omphalocele: Present	10	83.3	2	16.7	90	99	0.89	0.86	0.046
Absent	1	0.3	444	99.7					
Gastroschisis: Present	9	100.0	0	0.0	81	100	0.81	0.89	0.046
Absent	2	0.5	446	99.5					
SUA: Present	9	69.2	4	30.8	100	99	0.99	0.81	0.046
Absent	0	0.0	444	100.0					
Complex heart disease: Present	8	88.8	1	11.2	88	99	0.87	0.88	0.047
Absent	1	0.3	447	99.7					
Hydrops fetalis: Present	9	100.0	0	0.0	100	100	1.00	1.00	0.047
Absent	0	0.0	448	100.0					
Renal agenesis: Present	8	80.0	2	20.0	100	99	0.99	0.88	0.046
Absent	0	0.0	447	100.0					
Cardiomegaly: Present	8	72.7	3	27.3	100	99	0.99	0.83	0.046
Absent	0	0.0	446	100.0					
Cleft lips/palates Present	6	100.0	0	0.0	75	100	0.75	0.85	0.046
Absent	2	0.5	449	99.5					
Esophageal atresia: Present	7	58.3	5	41.7	100	98	0.98	0.73	0.045
Absent	0	0.0	445	100.0					

S: Sensitivity; E: Specificity; Y = Youden index, K = Kappa index; ONTD: Open neural tube defects; SUA: Single umbilical artery

Discussion

The literature describes a large variation in the frequency of congenital anomalies diagnosed during the prenatal and postnatal periods[6,20,21]. Current research indicates that the prevalence of fetal anomalies in populations of high-risk expectant mothers is around 27%[11]. The high frequency of malformations observed in our study (56.2%) is because the Fetal Medicine department at the I.M.I.P. receives screened cases from the whole state of Pernambuco and often from neighboring states. Furthermore, the exclusion criteria employed, such as births that did not take place at the I.M.I.P. and missing infant medical records, may also have affected these results.

A retrospective study in America with expectant mothers who underwent morphological ultrasound scans at between 15 and 26 weeks, found sensitivity and specificity of 71% and 99%, respectivelyx[22]. Another retrospective study, which assessed the effectiveness of prenatal ultrasonography for detecting congenital anomalies, reported specificity of 99.9%[23]. Although our study differed from some other published research by setting the lower limit of gestational age for morphological ultrasonography at the 22nd week, prenatal diagnosis of fetal anomalies achieved a sensitivity of 96% and a specificity of 79%. Another point that should be made clear is that our study recruited a smaller number of patients (n=457) when compared with other recent studies, which may explain the differences observed between the diagnostic validation figures.

The divergent results in the studies mentioned above are primarily the result of the study populations and the degree of specialization of the ultrasound professionals. While some of the studies are population-based, i.e. they cover all expectant mothers in a given period, others are carried out at a hospital level, including pregnancies at high-risk of congenital anomalies. One issue worthy of note is that some of these studies were carried out in maternity units with a primary level of complexity, and make unsatisfactory reference to high-risk patients.

Among these high-risk expectant mothers, morphological ultrasonography demonstrated good concordance with postnatal results. From a total of 287 patients with prenatal diagnoses of congenital anomalies, 42 were not confirmed after birth. According the European RADIUS study, ultrasonography should be performed at tertiary healthcare centers to allow for the best diagnostic investigation of fetal abnormalities, since Fetal Medicine specialists are better prepared to conduct fetal morphology studies than radiologists[11]. It should be emphasized that this study was undertaken at a center of excellence in Fetal Medicine by specialized professionals.

The literature describes the greatest frequency of prenatally diagnosed congenital anomalies occurring in the central nervous system, the genital and urinary systems and the musculoskeletal system[5,7,22] , whereas the greatest proportion of postnatal findings are in the circulatory system[7,24]. In our study it was observed that intrauterine diagnoses of central nervous system anomalies demonstrated excellent concordance with postnatal results, particularly ventricular dilatations and open neural tube defects. According to the literature, nervous system anomalies are most easily diagnosed during the prenatal period[24,25]. This is the result of the greater technical ease of obtaining this diagnosis, since, even when ultrasound scans are carried out by unspecialized professionals, measurement of the biparietal diameter and head circumference is an obligatory part of the examination. It should be stressed that, irrespective of operator ability, monitoring fetal growth encourages observation of intracranial structures that are neglected in routine scans[1,5].

There are few reports in the literature that provide data on sensitivity and specificity for specific fetal abnormalities in high-risk pregnancies. In a large proportion of studies, calculations are made on the basis of subdivision by major body system. For anomalies of the nervous system, sensitivity varies from 70% to 95%. In a prospective study with 3,685 fetuses presenting congenital anomalies, sensitivity for nervous system conditions was 88%[25]. Another study found a sensitivity of 76% among high-risk expectant mothers[22], and sensitivity of 93% has also been reported.[10] In our study, sensitivity for anomalies of this system was greater than the figures that can be found in the literature.

According to reports in the literature, anomalies of the genital and urinary tract, more specifically hydronephrosis, were diagnosed by antenatal ultrasonography in 1% to 5% of all pregnancies. Postnatal diagnostic confirmation varies with the severity of the defect. Just 11.9% of mild hydronephrosis or pyelocalyceal dilatations diagnosed with intrauterine ultrasound were confirmed postnatally, while 45.1% of moderate and 88.3% of severe hydronephroses were confirmed[23]. According to the Kappa index[13], prenatal hydronephrosis diagnoses had excellent concordance with postnatal findings. However, this concordance was not analyzed on the basis of the severity of dilatation. It should be pointed out that, for the purposes of genetic counseling, pyelocalyceal dilatation may be less traumatic for parents since in many cases there is no evidence of the anomaly after birth, which may be because of spontaneous regression of the defect or because the renal pelvis measurement had been overestimated by the ultrasound operator[23]. Fetal anomalies of the genital and urinary systems were detected with a sensitivity that is comparable with data from other available studies, where variation was from 69% to 94%[10,22]. Due consideration should be given to the fact that, in our study, ultrasound scans were carried out at a tertiary health center by Fetal Medicine specialists who are experienced at diagnosing fetal anomalies, in contrast with the majority of studies, which studied imaging examinations performed at basic healthcare centers by professionals who were not Fetal Medicine specialists[11].

The greatest number of discrepancies between prenatal and postnatal findings was related to musculoskeletal system anomalies (1.5%), which may have been caused by erroneous ultrasound diagnoses of certain defects because of confusion with postural defects or because results were compromised by changes in the volume of amniotic fluid (oligohydramnios), as is the case with congenital clubfoot and spinal column deformities.

Published data indicates lower sensitivity for malformations of the circulatory and musculoskeletal systems when compared with other systems and organs[10]. According to reports, diagnoses of cardiac fetal anomalies had sensitivity varying from 16% to 45% in a population study[10,22]. The 100% sensitivity in our study is in contrast to figures found in the literature, which could have been caused by the low number of expectant mothers who presented at the service already with a suspicion of congenital heart disease and, primarily, by the fact that the study was undertaken at a tertiary center where the health professionals have been trained in screening for cardiac anomalies. It should be emphasized that women with any type of family or obstetric history of congenital heart disease must be investigated more thoroughly and should also have morphological ultrasonography during the first trimester of the gestation, which itself offers sensitivity and specificity of 85% and 99%, respectively[26]. Other

studies have suggested sensitivity figures of 18% to 85% for anomalies of the musculoskeletal system[10,22]. At our service this sensitivity was similar to published data.

With respect to circulatory abnormalities, a certain difficulty can be observed in achieving intrauterine diagnoses, which may be caused by the low level of training of ultrasound operators in detecting anatomic and functional malformations of the fetal heart and also by the failure to investigate these anomalies systematically during routine obstetric ultrasound scans[8]. It has been found that around 25% of newborn infants leave the maternity unit without having heart disease diagnosed because many of them are asymptomatic at birth and only develop symptoms over the first 6 years of life[27]. With the introduction of fetal echocardiography, many diagnoses that had been missed by prenatal ultrasonography began to be detected. It is accepted that this test offers excellent diagnostic accuracy for describing the intracardiac anatomy, aiding postnatal treatment and prevention[28].

The diagnostic sensitivity results for anomalies of the digestive tract were not in line with figures from hospital-based studies (100% vs. 50% to 85%)[10,22]. It is believed that this difference may be related to the inclusion criterion of high or low-risk expectant mothers and with the timing and number of ultrasound scans in the several different studies found in the literature.

Overall, there is one methodological limitation that should be highlighted, which is that this was a retrospective study in which 532 cases were excluded. According to an earlier study carried out at our center using the same methodology, approximately 40% of cases were not included because of missing hospital records[29]. The importance of these exclusions is that the sensitivity and specificity calculations, while similar to the literature, may be under or overestimated. If this subset of cases had been primarily composed of "normal" cases that were then born with some type of abnormality, the method's rate of false-negatives would increase. Were there to be a significant proportion of abnormal ultrasound findings among the exclusions, it would be important to confirm the presence of these congenital anomalies since otherwise the rate of confirmed diagnoses reported could be under or overestimated.

Conclusions

Prenatal ultrasound diagnoses of congenital anomalies in high-risk pregnancies performed at specialist Fetal Medicine center had good concordance (K=0.76), validity (Y=0.75) and sensitivity when compared with postnatal results. Prenatal ultrasound detection of ventricular dilatation, neural tube defects, anencephaly, single umbilical artery, hydronephrosis, renal agenesis, clubfoot, cardiomegaly, complex heart disease, diaphragmatic hernia, omphalocele, gastroschisis, cleft lips/palates, ascites and hydrops fetalis all exhibited concordance with postnatal findings.

No conflicts of interest declared concerning the publication of this article.

References

1. Barini R, Stella JH, Ribeiro ST, Luiz FB, Isfer EF, Sanchez RC, et al. Desempenho da ultrassonografia pré-natal no diagnóstico de cromossomopatias fetais em serviço terciário. Rev Bras Ginecol Obstet. 2002;24:121-7.
2. Forrester MB, Merz RD. Genetic counseling utilization by families with offspring affected by birth defects, Hawaii, 1986-2003. Am J Med Genet A. 2007;143:1045-52.
3. Ceylaner G, Ceylaner S, Günyeli I, Ekici E, Celasun B, Danisman N. Evaluation of 2407 fetuses in a Turkish population. Prenat Diagn. 2007;27:800-7.
4. Nikkilä A, Rydhstroem H, Källén B, Jörgensen C. Ultrasound screening for fetal anomalies in southern Sweden: a population-based study. Acta Obstet Gynecol Scand. 2006;85:688-93.
5. Cecatti JG, Machado MRM, Krupa FG, Figueiredo PG, Pires HMB, et. al. Validação da curva normal de peso fetal estimado pela ultrassonografia para o diagnóstico do peso neonatal. Rev Bras Ginecol Obstet. 2003;25:5-40.
6. Chia SE, Shi LM, Chan OY, Chew SK, Foong BH. Parental occupations and other risk factors associated with nonchromosomal single, chromosomal single, and multiple birth defects: a population-based study in Singapore from 1994 to 1998. Am J Obstet Gynecol. 2003;188:425-33.
7. Carvalho VCP, Araújo TVB. Adequação da assistência pré-natal em gestantes atendidas em dois hospitais de referência para gravidez de alto risco do Sistema Único de Saúde, na cidade de Recife, Estado de Pernambuco. Rev Bras Saúde Matern Infant. 2007;7:309-17.
8. Amorim MMR, Vilela PC, Dutra Santos ARV, Lima ALMN, Melo EFP, Bernardes HF, et al. Impacto das malformações congênitas na mortalidade perinatal e neonatal em uma maternidade-escola do Recife. Rev Bras Saúde Matern Infant. 2006;6:519-25.
9. Stefos T, Plachouras N, Sotiriadis A, Papadimitriou D, Almoussa N, Navrozoglou I, et. al. Routine obstetrical ultrasound at 18-22 weeks: our experience on 7,236 fetuses. J Matern Fetal Med. 1999;8:64-9.
10. Lee RS, Cendron M, Kinnamon DD, Nguyen HT. Antenatal hydronephrosis as a predictor of postnatal outcome: a meta-analysis. Pediatrics. 2006;118:586-93.
11. Rankin J, Pattenden S, Abramsky L, Boyd P, Jordan H, Stone D, et al. Prevalence of congenital anomalies in five British regions, 1991-99. Arch Dis Child Fetal Neonatal Ed. 2005;90:F374-9.
12. Organização Mundial da Saúde (OMS). Classificação Estatística Internacional de Doenças e Problemas Relacionados à Saúde, Décima Revisão (CID 10). 8a ed. São Paulo: Editora da Universidade de São Paulo; 2000.
13. Pereira MG. Epidemiologia: teoria e prática. Rio de Janeiro: Guanabara Koogan; 2000.
14. Klein CH, Costa, EA. Os erros de classificação e os resultados de estudos epidemiológicos. Cad Saúde Pública. 1987;3:236-49.
15. Organização Pan-Americana de Saúde. Fundação Nacional de Saúde. Centro Nacional de Epidemiologia. Métodos de investigação epidemiológica em doenças transmissíveis; Rio de Janeiro; 1997.
16. Merz E, Welter C. 2D and 3D Ultrasound in the evaluation of normal and abnormal fetal anatomy in the second and third trimesters in a level III center. Ultraschall Med. 2005;26:9-16.
17. Rasiah SV, Publicover M, Ewer AK, Khan KS, Kilby MD, Zamora J. A systematic review of the accuracy of first-trimester ultrasound examination for detecting major congenital heart disease. Ultrasound Obstet Gynecol. 2006;28:110-6.
18. Eurenius K, Axelsson O, Cnattingius S, Eriksson L, Norsted T. Second trimester ultrasound screening performed by midwives; sensitivity for detection of fetal anomalies. Acta Obstet Gynecol Scand. 1999;78:98-104.
19. Goldberg JD. Routine screening for fetal anomalies: expectations. Obstet Gynecol Clin North Am. 2004;31:35-50.
20. Madi SA, Al-Naggar RL, Al-Awadi SA, Bastaki LA. Profile of major congenital malformations in neonates in Al-Jahra region of Kuwait. East Mediterr Health J. 2005;11:700-6.
21. Magriples U, Copel JÁ. Accurate detection of anomalies by routine ultrasonography in an indigent clinic population. Am J Obstet Gynecol. 1998;179:978-81.
22. Bricker L, Garcia J, Henderson J, Mugford M, Neilson J, Roberts T, Martin MA. Ultrasound screening in pregnancy: a systematic review of the clinical effectiveness, cost-effectiveness and womens views. Health Technology Assess. 2000;4:1-193.
23. França LC, Murta CGV, Moron AF, Montenegro CAB. Reflexão sobre a ultrassonografia na Obstetrícia: como melhorar a qualidade. Femina. 2004;32:167-70.
24. Benute GRG, Nomura RMY, Lucia MCS, Zugaib M. Interrupção da gestação após o diagnóstico de malformação fetal letal: aspectos emocionais. J Pediatr. 2003;79:10-7.
25. Aguiar MJB, Campos AS, Aguiar RALP, Lana AMA, Magalhães RL, Babeto LT. Defeitos de fechamento do tubo neural e fatores associados em recém-nascidos vivos e natimortos. J Pediatr. 2003;79:129-34.
26. Grandjean H, Larroque D, Levi S. Sensitivity of routine ultrasound screening of pregnancies in the Eurofetus database. The Eurofetus Team. Ann N Y Acad Sci. 1998;847:118-24.
27. Wren C, Reinhardt Z, Khawaja K. Twenty-year trends in diagnosis of life-threatening neonatal cardiovascular malformations. Arch Dis Child Fetal Neonatal. 2008;93:F33-5.
28. Gottliebson WM, Border WL, Franklin CM, Meyer RA, Michelfelder EC. Accuracy of fetal echocardiography: a cardiac segment-specific analysis. Ultrasound Obstet Gynecol. 2006;28:15-21.
29. Katz L, Amorim M, Coutinho I, Santos LC. Análise comparativa de testes diagnósticos para diabete gestacional. Rev Bras Ginecol Obstet. 2002;24:527-33.

Artigo recebido: 27/06/08
Aceito para publicação: 22/04/09