THE ROLE OF ULTRASOUND EXAMINATION IN DETECTION OF CONGENITAL ABNORMALITIES

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ABSTRACT

This is a prospective follow-up study involving 314 pregnant women who attended the U/S department in one of the main hospitals in Basrah city over the period from December 2003-December 2004.

The aim of the study was to assess the validity of ultrasound in prenatal detection of congenital anomalies in Basrah city. The majorities (64.3%) of pregnant women were attending for U/S examination in their third trimester, and only 0.3% were attending in the first trimester. The validity of prenatal U/S examination in detecting fetal congenital abnormalities was found to be very high. The study recommended that U/S examination should become part of the antenatal care provided in the primary health care centres.

INTRODUCTION

With the wide spread use of diagnostic U/S to evaluate the human fetus, it is now possible to diagnose number of anomalies of varying organ systems.[1] Ultrasound has become an invaluable tool in the antenatal diagnosis of many fetal abnormalities. A compilation of conditions which can be diagnosed antenataly listed 182 abnormalities and ultrasound was a major contributor to these diagnoses.[2] However, U/S as a mean of screening for fetal abnormalities has not been adequately assessed. A relatively recent research found that the fetal structures abnormalities including fetal skull, brain, spine, abdominal wall, limbs, stomach and bladder can be detected at 11-14 weeks scan in only 22.3 % of the cases; therefore, a second trimester anomaly scan was suggested in a routine antenatal care to increase the prenatal detection of the fetal defect. [3] However, some congenital abnormalities can not be detected by U/S prenatally but may be detected at birth (e.g. hare-lip) or shortly thereafter (e.g. congenital deafness, cataracts, congenital hypertrophic pyloric stenosis and renal tract abnormalities. [4] In Basrah no previous study was carried out to assess the validity of ultrasound examination in detecting congenital abnormalities during pregnancy. Such assessment is important as a prerequisite for the establishment of any screening programme. Thus the present study was carried out to measure the validity of ultrasound examination in prenatal detection of congenital anomalies in Basrah city.

METHODOLOGY

This is a prospective follow up study involving 314 pregnant women who were attending the ultrasound clinic in Al-Mawani hospital in Basrah city. The pregnant women were referred from the inpatient and outpatient departments of the same hospital over 12-month period extending between December 2003 and December 2004. All pregnant women who were attending on 2 selected days / per week who were examined by only two specialized radiologists were included in the study. The women were recruited into the study at the time of their attendance for U/S examination. They were informed about the study objectives and were included after giving a verbal consent to take part. None refused to participate, giving an overall response rate of 100%. A special questionnaire form was used for collection of data which included the following information: women's age, education, occupation, present obstetric history (parity, gravidity, last menstrual period, and any complication during the present pregnancy), and the reason for referral for U/S (indication of U/S). The data was collected by direct interview by one of the authors. The questionnaire form also included a section about the results of a single trans abdominal U/S examination which included the following: singleton or multiple, dead or alive fetus, gestational age, and presence or absence of congenital anomalies. The results of the ultrasound examination were derived directly from the ultrasound report. Then the studied pregnant women (64.3% in their third trimester, 35.4%in second trimester and only 0.3% in first trimester) were followed up until delivery to study the outcome of these pregnancies. The birth information requested included type of delivery, the fate of the fetus (alive or still

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birth), and the presence and type of congenital abnormality of the fetus. The sources of this information were: direct interviews with the women after delivery in their houses or at the clinic, or through a telephone call. Overall 23(7.3%) were lost to follow up. Analysis of the data was carried out using SPSS statistical package version 11. Chi-squared test was used for comparison. A P-value less than 0.05 was considered statistically significant.

RESULTS

The characteristics of the study population:

The study included 314 pregnant women, the majority were between 20-29 years old (58%) followed by those between 30-39 years of age (27.1%). Women who had primary schooling represented 39.5%, with only 16.6% of women were illiterate. The majorities (90.8%) were housewives with parity ranging between 0-3 (74.2%). Nearly half (49.7%) were referred for U/S examination for routine check-up; this was followed by attendance for ascertainment of gestational age of pregnancy. Only 8(2-6%) of pregnant women were referred for suspected congenital anomalies (Table-1). While the majority 259 (82.5%) were referred by a doctor, 55(17.5%) were self referred.

Results of U/S examination:

Based on U/S examination, out of the 314 pregnancies 10(3.2%) were multiple pregnancies, 57(18.2%) of pregnant women examined were carrying fetuses with congenital anomalies, and the majority (99.4%) of fetuses were alive.

Table 1. The reasons for referral to U/S examination.

Reasons for referral	No.	%
Routine check up	156	49.7
Gestational age	99	31.5
Suspected congenital anomaly	8	2.6
Fetal presentation	23	7.3
Others	28	8.9
Total	314	100.0

Types of congenital anomalies according to ultrasound examination:-

Types of congenital anomalies as detected by ultrasound are shown in (Table-2). The main types included the followings: hydrocephaly (36.9%) followed by anencephaly 18 (31.5%), Polycystic kidney (7.0%), and Encephalocele (5.2%).

Pregnancy outcome:

After delivery, 55 (18.9%) of the 291 pregnant women for whom the delivery information was available, gave birth to a child with one or more congenital anomalies. Fetal loss was reported by 34(11.7%) of the women (21 still births, and 13 abortions).

Table 2. Types of congenital anomalies according to U/S examination.

Congenital anomalies	No.	%
Anencephaly	18	31.5
Hydrocephaly	21	36.9
Omphalocele	1	1.8
Encephalocele	3	5.2
Polycystic kidney	4	7.0
Anencephaly + encephalocele	2	3.5
Microcephaly + encephalocele	2	3.5
Meningocele	2	3.5
Microcephaly	2	3.5
Hydrocephaly + absent limbs	1	1.8
Dwarfism	1	1.8
Total	57	100.0

Validity of U/S examination:

The majority of congenital anomalies which were diagnosed at birth were correctly detected by U/S examination during pregnancy (i.e. sensitivity of U/S was 98.2%). Only one case (1.8%) was missed by U/S. All negative cases by U/S didn't show obvious abnormalities at time of birth (i.e. specificity of U/S was 100%) (Table-3)

Table 3. Validity of U/S examination during pregnancy as a screening method for congenital anomalies.

Congenital anomalies by u/s	Congenital anomalies at birth				Total	
	+ ve		- ve			
	No.	%	No.	%	No.	%
+ ve	54	98.2	0	0.0	54	18.6
- ve	1	1.8	236	100.0	237	81.4
Total	55	100.0	236	100.0	291	100.0

$$\text{Sensitivity of U/S} = \frac{54}{55} \times 100 = 98.18\%$$

$$\text{Specificity of U/S} = \frac{236}{236} \times 100 = 100\%$$

DISCUSSION

Generally, significant abnormalities affect about 2% of births and mostly involve the central nervous system, urinary tract, limbs and heart.[2] The majority of anomalies occur in infants without family history of congenital malformations. So identification of anomalies provides parents with the opportunity for early counseling with the option of terminating pregnancy if a severe defect is detected. According to the present study the sensitivity of U/S examination as a screening test for congenital anomalies was 98% with a specificity of 100%. Only one birth with congenital blindness was missed. The results of a study carried out by an American group in 1996[5] found that the sensitivity of U/S done prior to 20 weeks gestation to detect anomalies ranged from 25% in physician offices to 71% at tertiary level hospital units. This discrepancy was attributed to differences in experience of the ultrasongrapher/ ultrasonologist, quality of equipment, and type of malformation being considered. A Spanish study of routine prenatal ultrasound screening for fetal abnormalities covering 22 years from 1970 to 1991 found that the overall detection rate of abnormalities under 22 weeks gestation increased in each 5 year period from 9% in 1970-74 to 85% in 1990-91.[6] Similarly, a Canadian review of the components of a complete second trimester ultrasound examination revealed that false positive rates of 0.2 to 1/1,000 women scanned were reported and that most initial false positive diagnoses were corrected on follow up evaluation.[7] The results of the present study showed a specificity of 100% with a false positive rate of zero which is in agreement with the Canadian studies. The high sensitivity of ultrasound examination as a screening test for congenital anomalies which was found in the present study may be partly explained by the fact that the examinations were carried out by two specialized, highly experienced ultrasonologists in a tertiary level hospital mainly during the third trimester. Furthermore, there is the possibility that many cases which are generally not recognized immediately after birth might have been missed. Although most anomalies of central nervous, gastrointestinal, genitourinary and skeletal systems are recognizable at birth, serious ones involving the cardiovascular system are often silent until the neonate demonstrate signs of cardiovascular compromise.[8] Therefore an actual measure of the validity of U/S examination during pregnancy would require a longer follow-up period and a proper examination of each infant by a specialist pediatrician rather than relying on parents' recognition and reporting of defects.

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