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The accuracy of ultrasound in the diagnosis of congenital abnormalities

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Original Article

The Accuracy of Ultrasound in the Diagnosis of Congenital Abnormalities

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Abstract

Objective: To determine the accuracy of ultrasound in the diagnosis of congenital abnormalities at the Aga Khan University Hospital, Karachi.

Methods: The data of congenital abnormalities was obtained from the obstetrical database and medical records of all cases complicated by congenital abnormalities, delivering from January 2001 to December 2003 and was reviewed. Antenatal ultrasounds had been performed by operators with different level of experience. In addition this data was retrieved from the termination and Congenital anomaly register. A structured data collection form was used to collect information of different variables of interest.

Results: Congenital abnormalities, complicated 2.8%(n=170), of all deliveries, including all cases of termination of pregnancy, stillbirth and live births. Out of the total, 11.6% occurred in women above the age of 35 years. Consanguinity was found in 18.2% cases. Prenatal diagnosis was made in just under half of the cases (48.8%). Central nervous system and renal abnormalities were commonly diagnosed. However, facial defects, heart defects or skeletal defects were more commonly missed.

Conclusion: Antenatal ultrasound successfully diagnosed foetal abnormalities in 48.8% of cases, and more than 90% Central Nervous system defects and renal abnormalities. In contrast about a quarter of Cardiac defects and none of the facial defects were detected. Based on these findings we recommend that the Sonologist should incorporate four chamber view of the heart and also look at the face carefully (JPMA 56:16;2006).

Introduction

Congenital anomalies occur in 2-3% of all births. They are an important cause of Perinatal morbidity and mortality and account for 20-30% of perinatal deaths.[1-4] Survivors have mental and physical disability. The psychological trauma and cost associated with foetal abnormalities, has lead to use of ultrasound for the prenatal diagnosis as an essential part of antenatal care.[4]

The diagnostic ability of ultrasound is well established by a number of studies.[5-7] Detection of foetal abnormalities depends on a number of factors including the nature or type of abnormality, sophistication of equipment and experience of operator. The Prevalence of abnormalities also depends upon the population being scanned. Therefore congenital abnormalities are higher among the referral center population as compared to the general population.[4]

In Pakistan where the social support system is virtually non-existent, bringing up a child with mental or physical handicap is a major burden for the parents and family. Primary prevention with Folic acid for this purpose has a limited role. In cases where primary prevention does not seem possible, prenatal diagnosis by ultrasound scan provides the next best alternative. The purpose of this study was to describe the trends of congenital abnormalities seen at a tertiary care facility in Karachi.

Table 1. Number of ultrasounds done by different operators.

Operator	Percentage	Number
Level three	22.4%	38
Level two	20.6%	35
Level one	33%	56
Others	24%	41
Total	100	

Material and Methods

This study presents the experience at the Aga Khan University Hospital, Karachi, Pakistan from January 2001-December 2003. This is a tertiary care teaching hospital of private sector in Karachi, equipped with latest diagnostic and therapeutic facilities. About a third of patients attending the department of Obstetrics and Gynaecology are high risk pregnancies. It is the practice in our department to offer two ultrasounds during pregnancy one before 12 weeks and the other at 20 weeks.

Congenital anomalies were defined as structural defects, chromosomal abnormalities, inborn error of metabolism or rare genetic syndromes, diagnosed either prior to or after birth. Minor abnormalities like hypospadias, skin tags, and low set ears have been excluded from the study.

Physicians performing ultrasound vary from those with limited experience doing a level scan to those experienced performing level three ultrasound. About a quarter of these examinations are also performed by Sonologists and Radiologists outside our University Hospital. In cases where abnormalities were diagnosed antenatally a repeat scan was performed at the Foetal Medicine Unit in the department of Obstetrics and Gynaecology.

All cases complicated by congenital abnormalities served as the study population. This data was gathered from various sources, that include congenital anomaly register, and hospital records of all live births and from the TOP, (Termination of pregnancy register) of all those undergoing termination of pregnancy for foetal abnormalities. Examination of the newborn/abortus comprised of clinical examinations, Radiological studies and chromosomal analysis if necessary.

The demographic detail of study subjects was noted. In addition, types of birth defect, sex and birth weight of the baby were also noted. All the variables were entered in a database file and analysed by using Statistical program for social sciences (SPSS version 10).

Table 2. Spectrum of abnormalities.

System Involved	Number	Percentage
Central Nervous System	36	21.2%
Cardiac defects	28	16.5%
Skeletal System	24	14.1%
Renal	20	11.8%
Facial defects	14	8.2%
Multiple abnormalities	14	8.2%
Miscellaneous	13	7.7%
GIT and Abdomen	10	5.9%
Down's Syndrome	5	2.9%
Meckel Gruber Syndrome	2	1.2%
Genitalia	4	2.3%
Total	170	100%

Results

Congenital abnormalities occurred among 2.8% of all deliveries. During the study period from January 2001-December 2003, a total of 8793 deliveries were reported. Out of these 170 cases of congenital abnormalities were identified and they served as the study population. These included terminations, live births and still births.

The mean age of the women in this study was 27.3 years with SD ± 5.3. Among the study subjects 11.6% were women above the age of 35 years. Only 8.8% of them had a previous history of congenital malformations. Consanguinity was found in 18.2% of cases. The mean gestational age at the time of ultrasound scan was 25.8 weeks (SD ± 6.8)

Ultrasound examination was performed by different operators. These ranged from those classed as level one to level 3. Forty percent of these ultrasounds were performed outside the hospital and for the purpose of analysis they have been put together in miscellaneous or others.

The ultrasound was able to diagnose congenital abnormalities in just under half of the cases (48.8%) whereas in 51.2% (83) malformations could not be diagnosed. The spectrum of abnormalities is shown in Table 2.

The most frequent abnormalities detected by ultrasound scan were of the kidney (19/20) followed by central nervous system (30/36). The details of abnormalities of the systems can be seen in Table 3.

Table 3. The accuracy of ultrasound for the individual systems.

System	Ultrasound diagnosed abnormalities/total number	Detection rate (Percentage)
Renal	19/20	98.8%
Central Nervous System	30/36	92.8%
GIT	6/9	33.3%
Multiple abnormalities	10/14	28.5%
Cardiac defects	7/28	25%
Skeletal System	4/24	20%
Down's Syndrome	None	0%
Facial defects	None	0%

Discussion

The current study evaluates various aspects of ultrasound screening at a teaching hospital in Karachi. Congenital abnormalities complicated about 2-3% of all pregnancies. This is consistent with that reported in the literature,[1-3] but higher than that reported by the EUROSCAN group.[4] This can be attributed to the high risk women seen in our population.

The sensitivity of ultrasound in the detection of foetal 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 sensitivity of the ultrasound scan for diagnosing congenital abnormalities in this study was 48.8%. Other studies have reported the sensitivity of ultrasound scan to be from 22-41%[8-10] to as high as 74-85%.[5-7] This can be due to the way these studies were conducted. Levi et al found in the earlier part of their study a lower sensitivity but later this improved as the technique and training improved.[9]

Consangiunity is also considered to be a risk factor for congenital abnormalities.[11] This association was not found in our study as consangiunity was found in only 18.2% of these cases.

The skill and experience of the sonographers is a critical factor in the detection of foetal anomalies.[3] As the ultrasound scans in this study were performed by people of varying experience, this can explain why approximately half of these abnormalities were missed on the examination in our study group.

The ultrasound scan failed to detect any facial defects in our study. Low prediction rate of 17.5% has been reported by some of the earlier studies.[12] However, more recent stduies claim the overall detection rate of as high as 65%.[13-15]

Similarly only a quarter of cardiac defects were diagnosed on the scan. The EURO scan study reported the sensitivity between 14-45%.[9] Eurenius et. al[10] has also reported similar results. The low detection rate was because the four chamber view was not included in the scan in their study population.

Based on our results we conclude that the antenatal ultrasound scan can be improved by incorporating four chamber views of the heart and careful look at the face during ultrasound examination. In addition, high risk women should be scanned in specialist units.

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