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Original Article

Diagnostic Accuracy of Ultrasonography in Detection of Fetal Neural Tube Defects in Polyhydramnios

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Objective: The objective of the study was to determine the sensitivity and specificity of ultrasonography in the diagnosis of fetal neural tube defects in cases of polyhydramnios.

Study Design: Descriptive type of study.

Place and duration of study: It was conducted in Radiology department of Mother and Child Hospital (MCH) in collaboration with gynaecology department of Mother and Child hospital (MCH), Pakistan Institute of Medical Sciences, Islamabad. Patients selected for study was both indoor and outpatient department cases referred for ultrasonography. Duration of study was one year.

Materials & Methods: In this study sixty (60) cases with the clinical suspicion of polyhydramnios were included. 3.5 Mega Hertz convex transducer was used for sonological assessment of these patients. Ultrasound findings were compared with the fetal outcome after delivery. Data was analyzed on SPSS 10.0 version and 2 x 2 tables were used to calculate test performance characteristic of Ultrasound finding.

Results: Total number of patients in our study was 60. Out of these 40 patients (66.6%) were ultrasound positive for neural tube defects which were confirmed by comparison with fetal outcome after delivery and considered true positive, while no patient was false positive. Out of these 18 (30.0%) were true negative while 2 (3.4%) were false negative, as confirmed subsequently by comparison with outcome after delivery.

The sensitivity, specificity, positive, negative predictive values and diagnostic accuracy of ultrsonography for neural tube defect in polyhydramnios was 95.2%, 100%, 100%, 90% and 96.6% respectively.

Conclusion: The sensitivity, specificity, positive predictive values and overall diagnostic efficacy of ultrasonography make it a modality of choice for detection of fetal neural tube defects in cases of polyhydramnios. It should be used as a primary screening investigation in patients with high clinical suspicion, since it is a safe, cost-effective, easy to perform and radiation free imaging technique.

Key Words: Neural tube defects, Ultrasonography, Polyhydramnios.

Introduction

Neural Tube Defects (NTDs) are due to failure of closure of neural tube in early embryogenesis. Pathophysiology of this common birth defect is due to abnormal process of primary and secondary neurulation. Resulting malformations are anencephaly, spina bifida, encephalocoele and meningomyelocoele.[1] Neural tube defects are associated with increased amount of amniotic fluid. Amniotic fluid index (AFI) more than 25cm or measurement of deepest single vertical pocket more than 8 cm is considered as polyhydramnios. [2] It is categorized in to mild, moderate and severe on the basis of amniotic fluid index. In mild it is 25-29.9cm, in moderate (30-34.9cm), in severe more than 35 cm. [3]

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Sonographic estimation of amount of liquor is very important parameter in antenatal Surveillance. [4,5] Sonographically polyhydramnios can be diagnosed at 12 to 13 weeks. But more reliable diagnosis is at 15 to 16weeks.[6] Alteration in amount of liquor indicates fetal compromise and is suggestive of target imaging for fetal anomalies. [7,8]

With increasing severity of polyhydramnios there is increase risk for fetal anomalies. [9] Anomalies associated with polyhydramnios are NTD, midline facial defects, cardiac septal defects and tracheoesophageal fistula. [2.] Ultrasonography (US) is primary and most frequently used diagnostic tool for antenatal diagnosis of fetal anomalies. In routine anomaly scanning is done after 20 weeks. [10] Other important pathological investigation is level of maternal serum alfa fetoprotein at 18 to 20 weeks. Alfa fetoprotein is found in both fetal serum

and amniotic fluid. It is produced early in gestation by fetal yolk sac and later by liver and gastrointestinal tract. High level of AFP suggest that developing fetus may have neural tube defect like spina bifida, anencephaly . It may also represent failure of closure of fetal abdominal wall.

The purpose of study is assessment of accuracy of US in diagnosis of fetal neural tube defects in cases of polyhydramnios. It is easily available, cost effective, easy to perform and safe. There is no risk of radiation exposure to fetus. This will help patient for early intervention in cases of anomalies.

Gold standard in this study was be fetal outcome after delivery, ultrasound finding regarding fetal neural tube defects in polyhydramnios were compared with gold standard and accuracy determined by calculating sensitivity, specificity, positive predictive value, negative predictive value with help of 2x2 table. Our aims and objectives were to determine sensitivity and specificity of US for detection of fetal NTD in cases of polyhydramnios.

Material and Methods

All the indoor and OPD/booked and non-booked pregnant women presenting at the Radiology Department of Mother and Child Health (MCH), Pakistan Institute of Medical Sciences, Islamabad were screened for enrolment. Those who fulfilled the below mentioned inclusion criteria were enrolled in this study.

The total duration of the study was chosen as one year. The enrolment of patients was started on 21 .03.08 and data were completed by 20.03.09. Sixty patients were enrolled during study period of one year.

The purposive (non-probability) sampling technique was used.

The pregnant women who fulfilled the following inclusion criteria were enrolled:

- Pregnant women of age group 15 to 35 years, though according to World Health Organization (WHO) reproductive age is 15 to 49 years but above 35 years there is increased risk of anomalies.
- Amniotic Fluid Index (AFI) was more than 25 cm.
- Pregnant women with gestational amenorrhea of more than 20 weeks.
- Informed consent.

The pregnant women who had any of the following exclusion criteria were not enrolled. These were:

- Pregnant women in which follow up is not possible.
- Anomalies other than neural tube defect.
- Already diagnosed cases.
- Pregnant women without consent.
- Any other co-morbid condition such as diabetes.

All the information recorded in the Performa was computerized by using the software SPSS version

Results

Total number of patients in our study was 60. Out of these 40 patients (66.6%) were US positive for NTD which were confirmed by comparison with fetal outcome after delivery and considered true positive, while no patient was false positive. Out of these 18 (30.0%) were true negative while 2 (3.4%) were false negative, as confirmed subsequently by comparison with outcome after delivery. The sensitivity, specificity, positive, negative predictive values and diagnostic accuracy of US for NTD in polyhydramnios was 95.2%, 100%, 100%,90% and 96.6% respectively.

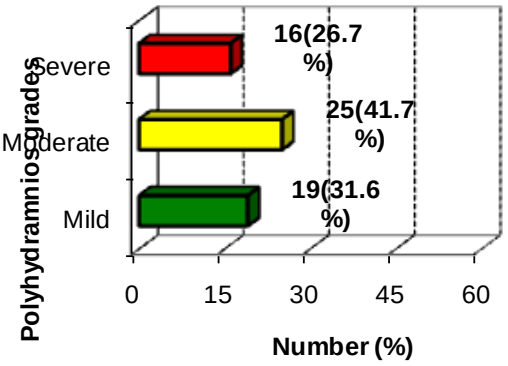

Figure 1: Distribution of grades of polyhydramnios among all the enrolled women (n = 60)

Table 1: Distribution of types of fetal anomalies detected by ultrasonography among women detected to have fetal anomalies

Type of fetal anomalies	Number of patients	Percentage
Anencephaly	25	62.5%
Spina Bifida	12	30.0%
Meningomyelocoele/Encephalocoele	3	7.5%

Table: 2 Comparison of results of ultrasonography with fetal outcomes after delivery

US Findings	Final Diagnosis	
Test Result	Diseased	Not Diseased
US Positive 40	True Positive 40	False Positive 0
US Negative 20	False Negative 2	True Negative 18

Table 3: Sensitivity, Specificity, positive predictive, negative predictive value and diagnostic accuracy of ultrasonography in detection of fetal anomalies among all the enrolled women (n = 60)

Sensitivity	95.2%
Specificity	100.0%
Positive predictive value	100.0%
Negative predictive value	90.0%
Diagnostic accuracy	96.6%

Figure 2: Deepest pocket of liquor in polyhydramnios.

Figure 3: Anencephaly with frog eye appearance.

Discussion

Ultrasonography is important, non invasive, safe, easily available and cost effective technique for prenatal diagnosis of amount of liquor and neural tube defects [11]. Prenatal diagnosis is helpful for patient as well as for obstetrician because it enables timely medical or surgical treatment of patient before and after birth. Secondly, it gives chance to parents to prepare them psychologically, socially, financially and medically for a baby with health problem. Thirdly it gives chance to parents for early termination with diagnosed condition.

In our study 60 patients were selected who were sent to radiology department for routine ultrasonography or with suspicion of polyhydramnios, regardless of their race, religion and socioeconomic status. Brief clinical history was taken about gestational age, previous pregnancies and expected date of delivery from the patient. The ultrasound was done after informed consent. The results of ultrasound were compared with fetal outcome after delivery.

The mean (standard deviation) age of all the enrolled women at the time of presentation in our study was 28.6 (± 4.6) years with median of 29 years. The youngest pregnant woman was 18 years old, whereas the oldest woman was 35 years of age. Majority of women were between the age group 26 to 30 years as 30 (50.0%) were of this age group. Thirteen (21.7%) pregnant women in our study were up to the age of 25 years, whereas 17 (28.3%) were older than 30 years of age. In another study which was done by Scheltema AVPNA et al[12] in 6 years period .In their study 4470 patients were screened for CNS anomalies. Mean maternal age in that study group was 30 years. [12]

The grades of polyhydramnios were calculated according to AFI and patients were categorized in to mild moderate and severe forms. Among these patients nineteen (31.7%) had mild polyhydramnios, 25 (41.7%) had moderate and 16 (26.7%) had severe polyhydramnios.

In our study the gestational age at time of examination was 25.8 weeks with median of 26 weeks. Gestational age between 18 to 28 weeks is considered ideal time for anomaly scan because high quality sonographic images are available at this time. It has been reported by Hegge FN et al that in 2/3rd of prenatally diagnosed fetal anomalies clinical sign and symptoms may develop at 23 to 24 weeks of gestation. [13]

In this study according to socio economic status the distribution is as 29(48.3%) belonged to low, 28 (46.7%) belonged to middle and only 3(5%) belonged to high socioeconomic class. Therefore according to our study the prevalence of NTD is most common in low socioeconomic group. This can be explained due to dietary factors and lack of awareness in lower class. Role of folic acid in preconception and in first trimester is now gaining significance in prevention of NTD. Morris JK et al and Kadir RA et al reported that the incidence of live births, stillbirths and pregnancies terminated because of a fetal neural tube defect has fallen steadily from 1972 to 1992 due to folic acid therapy. [14, 15]

In our study 40 patients were true positive, 18 were true negative, 2 were false negative and there was no

false positive. 2 false negative were those in which ultrasound showed no anomaly while fetal outcome after delivery turned them in spina bifida.

Filly RA et al reported that spina bifida among NTD is most difficult to detect by ultrasound. Such anomalies are sonographically manifested by splaying of the posterior ossification centers of the spine.[16] The transverse plane has been reported as the most useful plane for scanning these defects[16].In another study Fiske CE et al guide that meticulous scanning is necessary, as a slight widening of the interpedicular distance is a normal finding in the cervical and lumbar areas. [17] Thus these two studies reported in literature support false negative cases of our study

Sensitivity, specificity, PPV, NPV and diagnostic accuracy in our study are 95.2%,100%,100%,90%and 96.6% respectively .In study done by Vos JM et al sensitivity of ultrasound for spina bifida was about 71% and a specificity close to 100%.[18] In another study of same category done by Brand IR et al on 2261 cases specificity of ultrasound for detection of fetal anomalies was 99.5%.[19] Similarly study done by Antsaklis AJ showed specificity of ultrasound 99.9%.[20] So it can be seen clearly that our results match well with those of previous studies reported in literature.

Conclusion

Ultrasound when performed under experience hands, is the most accurate method of detecting neural tube defects in the second trimester of pregnancy.

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