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Original Research Article

Failure of Ultrasound in Prenatal Diagnosis of Neural Tube Defects in Rural Sindh, Pakistan

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ABSTRACT

Introduction: Neural tube defects (NTDs) are the leading cause of children mortality and morbidity around the globe. This study is designed to observe the efficacy of ultrasound in prenatal diagnosis of NTDs in our setup.

Methods: This descriptive study was conducted in Department of neurosurgery at Liaquat University hospital Jamshoro with collaboration of Department of Anatomy of Liaquat University of Medical and Health Sciences from September 2013 to December 2013. A total 45 patients were studied in this study. A detailed history for any risk factor, family history of NTDs along with biodata was taken and recorded on a proforma of all patients. A thorough physical examination was performed, type of neural tube defect noted. Ultrasound reports of antenatal checkups which were available were thoroughly evaluated to examine the failure of ultrasound in prenatal diagnosis of NTDs in our setup.

Results: Total 45 patients with varying age groups ranging between 01 day old and 05 years old were examined, out of which 18 were male (37%) and 27 (62%) were female patients. Only 09 out of 45 cases of (20%) NTDs were diagnosed before birth while rests were (80%) diagnosed after birth. In diagnosed cases 02 were diagnosed at rural and 07 at urban, while in undiagnosed cases 30 at rural and 06 at urban health care centers

Conclusions: Antenatal ultrasound is a non–invasive, highly sensitive, accurate imaging technique which gives good results in experienced hands. Proper utilization of this investigation may help in reducing the burden of handicapped children and decreasing the mortality rate due to NTDs.

Key Words: Folic acid, Health care, NTDs, Rural, Ultrasound.

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INTRODUCTION

Neural tube defects (NTDs) are the leading cause of children mortality and morbidity around the globe. It affects around 4 to 15 per 10,000 live births. Its frequency varies according to ethnicity, geographic location and environmental factors. The precise reason behind these defects remains unidentified. Various etiological factors such as combination of genetic predisposition, nutritional and environmental causes, female gender, family history of NTDs, obesity, maternal pregestational and gestational diabetes, low dietary folate intake, lack of folic acid supplementation, use of anticonvulsant drugs (sodium valporate, carbamazepine), use of folic acid antagonists (e.g. methotrexate), might be responsible for failure of neural tube closure leading to NTDs[1,2].

One pregnancy with NTDs carries increased risk of NTDs in subsequent pregnancies. It is reported in previous studies that the ratio of couples having one previous baby with NTDs is 1 in 33 while with two past pregnancies this ratio increased to 1 in 10. With advances in prenatal diagnostic tools for congenital anomalies, NTDs are the easiest among all birth defects to diagnose prenatally. First trimester scanning detect >90% cases of anencephaly, 80% of encephalocele and 44% spina bifida. Second trimester scan for fetal anomaly detect 98% anencphaly and 90% open Spina bifida. Gestational age influences the type of NTD diagnosed.[3,4]

Prenatal detection of an NTD helps the parents and health practitioners to manage the problem more effectively after evaluation of the anomaly and overall health status of the fetus. An extreme form of NTD such as anencephaly incompatible with life needs termination of pregnancy. A caesarean delivery is preferred in some NTDs like spinal meningomyelocoele to avoid more neurological deficit in new born. Antenatal repair of NTDs is being performed in highly advanced tertiary care hospitals in developed countries for selected cases.[5]

With the introduction of screening methods for detection of NTDs in 1970, an increase in total number of reported cases was observed. However, overall impact was a decreased prevalence of NTDs at birth. This decline strongly suggests the importance of early antenatal diagnosis of NTDs followed by medical termination of pregnancy (MTP). Furthermore folate prophylaxis is also an essential factor for reduction of NTD incidences. Although some factors remain unalterable such as consanguinity and racial variables of the parents.[6,7]

Ideal protocol for prenatal screening of NTDs and their diagnosis is universally offered in second trimester between 15 and 22 weeks' gestation, for accuracy and optimal management of affected pregnancies. The accuracy of diagnosing these defects is effected by certain factors such as type of NTD, gestational age, maternal weight, maternal insulin-dependent diabetes, multiple gestations, ethnicity, environmental factors (prescribed and non-prescribed medicines), and multiple fetal anomalies.[8,9]

An ideal prenatal screening tool must meet the criteria of acceptability, validity, simplicity, safety, rapidity, ease of administration and cost. It must be highly specific and sensitive. Prenatal diagnosis techniques include:

- Assessment of serum markers such as maternal serum alpha-foetoprotein and acetylcholinesterase activity;
- Prenatal ultrasonography, Fetal magnetic resonance imaging (MRI)
- Amniocentesis.

Out of all these techniques ultrasound is gold standard diagnostic tool with similar sensitivity and lower false positivity compared to serum markers. Each laboratory has its own normative data standardized for different durations of gestation so inter laboratory standard for serum markers is difficult to assess. MRI is cost consuming, expert dependent, not available with ease and nor at all in remote areas, similar is with amniocentesis with 0.5% chances of miscarriage; keep these techniques less applicable than ultrasound.[1]

Prenatal ultrasonography is primary, simple and widely available non-invasive screening modality of choice for the detection of fetal anomalies. It is advised routinely during second trimester in all pregnancies. Modern high-resolution ultrasound machines have unique potential in evaluating NTDs at early stages of development. The most common defects such as anencephaly and encephalocele are easily visualized on ultrasound while identification of spinal deformities such as meningomyelocoele is difficult to diagnose and needs expertise. Ultrasound technology has improved remarkably nowadays with special expertise in obstetric ultrasonography report, excellent sensitivity and specificity in detecting fetal neural tube defects.

In the hand of experienced operators, ultrasonography alone has up to 97% sensitivity and 100% specificity in the diagnosis of NTDs. In less experienced hands, however, ultrasonography is only a screening test that can have a high false-negative rate.[10,11]

Despite of several screening tests available for prenatal detection of NTDs, standardized protocol is still lacking in developing countries like us. These anomalies can be diagnosed with reasonable accuracy in antenatal period and can be prevented to a large extent, by avoidance of known teratogens, pre and peri conceptional use of folate.

This study is designed to observe the efficacy of ultrasound in prenatal diagnosis of NTDs in our setup as majority of our patients referred to tertiary care hospital from rural/remote areas of Sindh deprived of basic health facilities

METHODOLOGY

This descriptive study was conducted in Department of neurosurgery at Liaquat University hospital Jamshoro with collaboration of Department of Anatomy of Liaquat University of Medical and Health Sciences from September 2013 to December 2013. A total 45 patients were studied in this study. All are admitted throughout patient department. A detailed history for any risk factor, family history of NTDs along with biodata was taken and recorded on a proforma. A thorough physical examination was performed, type of neural tube defect noted. Ultrasound reports of antenatal checkups which were available were thoroughly evaluated to examine the failure of ultrasound in prenatal diagnosis of NTDs in our setup.

Table 1: Age and Gender of Patients (n=45)

Age	Gender	
	Male	**Female**
Neonate	02	08
Infant	10	13
>1 year	05	07
	17 (37%)	28(62%)

Table 2: No. of cases diagnosed on ultrasound before and after birth in different demographic locations

Diagnosis on antenatal u/sound before birth	
Rural	Urban
02	07
Total = 09(20%)	
Diagnosis on antenatal u/sound after birth	
Rural	Urban
30	06
Total = 36(80%)	

Table 3: No. of ultrasound in patients

	No. of ultrasound in patients of NTDs						
Diagnosed after birth	**Nil**	**01**	**02**	**03**	**04**	**05**	**Total No.**
	08	09	04	08	05	02	36
Diagnosed before birth	00	03	01	02	03	00	09

RESULTS

During study period total 45 patients with varying age groups ranging between 01 day old and 05 years old were examined, out of which 18 were male and 27 were female patients. On physical examination type of anomaly was observed. Available ultrasound reports were thoroughly reviewed with other details. Following Tables show the observations made during the study.

DISCUSSION

The babies born with congenital anomalies are a cause of mental distress and economic burden on parents. Approximately 2-3 per 100 children are born with some kind of birth defects around the globe every year. Out of all anomalies 2.5/1000 babies are born with NTDs. These defects are the major cause of morbidity and mortality in new born.[12,13]

There is plenty of data available on prenatal diagnosis of NTDs throughout world including Pakistan but this is the first study conducted at LUMHS to highlight the problem of antenatal non diagnosis of NTDs in rural areas of Sindh Pakistan. In particular this study was intended to get an idea about the failure of ultrasound in diagnosis of different NTDs in antenatal period.

The results demonstrate the higher incidence of failure of NTDs diagnosed in antenatal period. One reason might be due to the fact that the majority of patients residing in remote areas of Sindh which are deprived of basic health facilities. Where the basic health units are available, they are either not provided with proper equipment/machines such as ultrasound machine, or lack of experts in the field. In the absence of expert doctor or sonographer poor patients rely on mercy of lady health workers or technicians who are unable to provide health care up to mark.

Moreover, the population in these rural areas belongs to low socioeconomic status, with lack of education and lack of health care awareness. Different myths is another serious issue of subcontinent which hinders there ways to seek medical advice and care during pregnancy.

The incidence of different NTDs varies according the geographic conditions, race and gender of baby. It is twice common in females than males, like others same ratio is seen in this study also which show 28 (62%) female in 45 cases compared with 17 (37%) males.[14]

The results of this study demonstrate that only 09 out of 45 cases of (20%) NTDs were diagnosed before birth while rests were (80%) diagnosed after birth. In diagnosed cases 02 were diagnosed at rural and 07 at urban, while in undiagnosed cases 30 at rural and 06 at urban health care centers. These results show medical negligence contrary to medical ethics but it may be the result of deprived health facilities, furthermore one reason is the patient's awareness for seeking medical care as shown in table 3 in which 08 cases never had ultrasound or antenatal checkup therefore NTD in them diagnosed after birth.

The diagnostic ability of ultrasound is well established by a number of studies. Detection of fetal abnormalities depends on a number of factors including the nature or type of abnormality, sophistication of equipment and experience of operator.[15,16]

In Pakistan where poverty and illiteracy are prime factors to hinder primary prevention with folic acid, prenatal diagnosis with ultrasound is the second best alternative to prevent the birth of handicapped child. As bringing up a child with mental or physical abnormality is a major burden for the parents, family and society as whole. Ultrasound offers the possibility of early and accurate diagnosis of neural tube defects, and has therefore improved medical counseling and parents' decision making.[17,18]Folic acid supplementation reduces the risk of NTD by 35-70% but a non-declining birth prevalence of NTD is a concern to our country like other developing countries. Possible causes of failures may be insufficient recommendations, non-compliance of mothers, folic acid fortification of food and myths.[19-21]

CONCLUSION

Prenatal diagnosis has improved during the last 30 years but due to lack of awareness and economy still not practiced in developing countries like Pakistan. Proper preconception counseling and awareness is required which is currently lacking in remote areas of Pakistan. Antenatal ultrasound is a noninvasive highly sensitive, accurate and cost effective imaging technique which gives good results in experienced hands. Proper utilization of this investigation may help in reducing the burden of handicapped children and also helps in reducing the mortality rate due to NTDs.

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