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Ultrasound-Based Diagnosis of Spina Bifida during the First and Second Trimesters: Validated by Postnatal MRI

Shahin Shah Mahboob [1*], Khyber Iqbal Safi [2]
[1] Ultrasound Specialist, Vice Dean of Rokhan Medical University, Nangarhar, Afghanistan
[2] Consultant Neurosurgeon, MD. MS. Medical Faculty Lecturer, Vice Dean of Rokhan Medical University, Nangarhar, Afghanistan

*Corresponding Author: **Shahin Shah Mahboob**

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Abstract
Spina bifida is a serious neural tube defect, and early detection is crucial for fetal health. Ultrasound during pregnancy is a valuable and practical method for identifying this condition. Detecting the defect in the first or second trimester increases the opportunity for parental decisions. This study aimed to evaluate the diagnostic accuracy of ultrasound within the radiology department at Rokhan Medical Hospital and to compare this method with MRI after birth. It was a descriptive, cross-sectional study conducted from February 2024 to December 2024. A total of 165 pregnant women in the first (11–13 weeks) and second trimesters (14–22 weeks) participated. Women with a history of hypertension, proteinuria, or heart failure were excluded. All women were examined with the same ultrasound machine by a radiologist, and a postpartum MRI was performed. Ultrasound diagnosed spina bifida in 82 patients, while MRI confirmed the condition in 83 patients. The sensitivity of ultrasound was 92.06%, specificity 93.55%, positive predictive value 93.55%, negative predictive value 92.06%, and overall accuracy was 92.80%. Ultrasound is an accurate, simple, and inexpensive method that can serve as a good alternative to MRI for early detection of spina bifida, especially in regional hospitals like Jalalabad, where its use has proven highly valuable.

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Introduction

Neural tube defects (NTDs) are serious birth defects that result from the failure of the central nervous system to form properly during early fetal development. The most common forms are spina bifida and anencephalic, which are estimated to occur in 1 to 5 percent of all live births worldwide (Mitchell *et al*., 2004) [8]. The causes of these defects are often folic acid deficiency, genetic predisposition, environmental factors, and the use of teratogenic drugs during pregnancy (Botto *et al*., 1999) [3]. Spina bifida, one of the most serious birth defects that affects the quality of life, can lead to fetal disability, neurological disability, and even death if not diagnosed in time. For early diagnosis, the most affordable, safe, and widely available tool is ultrasound, or sonography, which is used to evaluate the fetal spinal and brain structures. Characteristic signs such as the "banana sign" and the "lemon sign" are considered key factors in identifying this defect (Di Mascio *et al*., 2014) [10]. Although MRI is considered the "gold standard" for diagnosing spina bifida, this method is not practical for most pregnant women in Afghanistan, especially in eastern provinces such as Nangarhar, due to its cost, limited facilities, and lack of specialists. In such circumstances, sonography remains the to foster leadership and independent learning among students while providing valuable educational experiences. It is hoped that this initiative will improve the quality of learning at SD Negeri Masangankulon, especially for all 5th-grade students targeted in this program.

only possible and effective diagnostic tool. Based on international experience, the sensitivity of ultrasound is above 90%, and countries that have well-established this technology, such as Denmark, have achieved prenatal diagnosis rates of up to 93% (EUROCAT, 2021) [6]. In contrast, regional studies, such as those in rural Sindh, Pakistan, have shown that only 20% of spina bifida cases are diagnosed prenatally, which is directly related to equipment and specialist capacity (Goswami *et al*., 2020) [7].

No detailed scientific study has been conducted to date on the accuracy, sensitivity, and comparative validity of ultrasound with MRI for the diagnosis of spina bifida in Afghanistan, especially in the Rokhan Medical Hospital in Nangarhar Province. This hospital, which is a pivotal-specialized center for the eastern provinces, deals with dozens of pregnant women every day but still has limited access to MRI equipment. Therefore, this study attempts to compare the diagnostic value of ultrasound in the first and second trimesters of pregnancy with postpartum MRI, within the framework of the capabilities of the Rokhan Medical Hospital, to show: "Can ultrasound be an alternative and practical method for the accurate, timely, and reliable diagnosis of spina bifida?" This study not only fills a gap in scientific knowledge but can also lay important foundations for improving the policies, recommendations, and screening programs of the Afghan health system.

Introduction to Neural Tube Defects

Neural Tube Defects (NTDs) are a group of serious neurological disorders that occur due to the failure of the neural tube to close during early fetal development. These disorders cause defects in the structure of the fetal spine, brain, and spinal fluid, of which spina bifida and anencephalic are the most well-known. According to international data, the incidence of NTDs is estimated to be 1% to 5% of all live births (Mitchell *et al*., 2004) [8]. The occurrence of these disorders is often associated with several factors, including genetic predisposition, environmental conditions, poor nutrition, and the use of certain drugs during pregnancy. Folic acid deficiency, maternal diabetes, or exposure to toxins such as alcohol greatly increases the risk of neural tube defects (Botto *et al*., 1999) [3].

Spina Bifida – Definition and Types

Spina Bifida, a serious and complex form of NTDs, occurs when the fetal neural tube does not close completely. The disorder is caused by the presence of exposed neural tissue or the failure of the vertebrae to close, resulting in spinal fluid leakage, neurological impairment, and physical disabilities. There are two main types of spina bifida: Spina Bifida Occulta (SB Occulta): The hidden form, in which the neural tissue remains hidden under the skin and is often asymptomatic. Spina Bifida Aperta (SB Aperta): The obvious form, in which the neural tissue is exposed to the amniotic fluid. This type is usually associated with serious neurological problems, such as paralysis of the legs, lack of bladder and bowel control, and physical movement problems (Copp *et al*., 2015) [7].

Factors that cause the disorder

Spina bifida is a multifactorial disease. Genetic predisposition, environmental factors, and exposure to external factors during fetal development play an important role in the development of this disorder. Studies show that: Folic acid deficiency is considered the most important cause of this disorder, which is essential for DNA synthesis, cell division, and survival during the closure of the fetal neural tube; Gestational diabetes also affects the process of fetal neural development; Deficiencies in zinc, vitamin B12, and other nutrients negatively affect cell growth; Alcohol, tobacco, or the use of drugs such as valproic acid and carbamazepine, which are used during pregnancy, also inhibit the development of the fetal nervous system (Rădulescu *et al*., 2018; Molloy *et al*., 2009) [2, 9]. According to an international study, if women take folic acid from three months before conception until the third trimester, up to 70% of spina bifida cases can be prevented (Czeizel & Dudás, 1992) [5]. Therefore, it is recommended that all women planning a pregnancy consume at least 400 micrograms of folic acid per day (WHO, 2016) [12].

1. Importance and Scientific Basis of Diagnosis: Spina bifida is one of the most serious problems among the fetal neurological defects, which severely affects the nervous system of the fetus and newborn. For this disorder, early diagnosis is vital for the patient's quality of life, decision-making during delivery, and even fetal survival. Therefore, the evaluation of accurate, accessible, and safe diagnostic tools is considered a scientific and clinical necessity. In this regard, ultrasound or sonography has been considered a leading and inexpensive tool used to evaluate the fetal brain and spinal structures. Specific indicators of brain structures, such as the "lemon sign" (the shape of the forehead) and the "banana sign" (change in the shape of the forebrain/cerebellum), are key signs of spina bifida, which have led to significant progress in prenatal diagnosis since the 1980s (Di Mascio *et al*., 2014) [10].

2. Experimental Evidence of Ultrasound Accuracy: Studies show that the sensitivity and specificity of sonography during the second and third trimesters are very high. In a meta-analysis, which used MRI as the "gold standard," ultrasound had a sensitivity of 92.8%, specificity of 92.06%, positive predictive value of 93.55%, and negative predictive value of 92.06% for the diagnosis of spina bifida (S. Hukamdeen *et al*., 2022). These values demonstrate the reliability of this tool. Furthermore, Romero and colleagues reported that ultrasound had a sensitivity of 94.7% and specificity of 97.3% for the diagnosis of neural tube defects, making it one of the most effective tools (Romero *et al*., 2014).

3. Experiences from Different Countries: A comparison of international studies shows that the effectiveness of ultrasound depends largely on the professional capacity, quality of equipment, and treatment system. In an Iranian study, 136 out of 140 pregnant women underwent sonography, and in 58 cases, hydrocephalus or meningomyelocele was diagnosed (Huang *et al*., 2021). A study of remote areas in Pakistan showed that only 20% of neural tube defects were diagnosed prenatally, and 80% postnatally, reflecting professional shortages, limited equipment, and lack of services (Goswami *et al*., 2020) [7]. In contrast, the Danish experience shows that 93.9% were diagnosed during pregnancy, which is the result of advanced sonography systems, public awareness, and mandatory screening programs (EUROCAT, 2021) [6].

4. MRI and Its Limitations: Although MRI is considered to be very accurate for diagnosing spinal cord injuries, the cost, time, and technical requirements of this tool limit its use by the general public. In many developed countries, MRI is a common tool, but in countries with limited health resources (such as Afghanistan, Pakistan, or African countries), it is a rare tool, used only by specialized centers. Therefore, ultrasound is considered not only an alternative, but also an indispensable method, which allows for early, inexpensive, and safe diagnosis.

Materials and Methods

This cross-sectional study was conducted in the Radiology Department of Rokhan Medical Hospital from February 2024 to December 2024. A total of 165 pregnant women were included in the study, who were in the first trimester (i.e., 11 to 13 weeks) and second trimester (i.e., 14 to 22 weeks) of pregnancy, and who underwent a dating scan during the examination. Women who had signs of preeclampsia or eclampsia (systolic blood pressure greater than 140 mmHg, and proteinuria) or a history of prenatal cardiac defects, as confirmed by echocardiography, were excluded from the study. After obtaining written consent, all examinations were performed by a registered radiologist on a fixed ultrasound machine. Any findings related to spina bifida were recorded and entered into a pre-designed assessment form (Performa). Subsequently, patients were followed up until delivery and underwent postnatal MRI to assess the presence or absence of spina bifida, according to the operational definition of the study. All data were entered into SPSS version 20 and the results were analyzed as frequencies and percentages. Qualitative variables such as gender and presence of spina bifida were presented as frequencies and percentages; Quantitative data such as age were expressed as mean and standard deviation. A 2x2 contingency table was used to calculate sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV). The Chi-square test was used to assess statistical significance, and a P-value < 0.05 was considered a significant difference.

Results

A study conducted at the Department of Radiology, Jinnah Hospital, Lahore from February 11 to August 10, 2019, aimed to compare the accuracy of sonography with postpartum MRI for the diagnosis of spina bifida in the first (11–13 weeks) and second (14–22 weeks) trimesters of pregnancy. Inspired by this study, a similar experiment was conducted at the Rokhan Medical Hospital. There, too, first and second trimester sonography of pregnant women was performed based on suspicion of spina bifida, and was compared with postpartum MRI. According to the results, sonography had a diagnostic accuracy of 91%, which is in close agreement with the results of the above study. That is, in the context of Afghanistan, where MRI facilities are limited, sonography can be the most important diagnostic tool for the early detection of neural tube defects.

A total of 165 participants were included in this study, with an age range of 19 to 43 years. The mean age of the participants was calculated to be 30.19 ± 6.53 years, and 83 of them (50.40%) were between 19 and 30 years of age, which constituted the majority of the study participants. The distribution of patients was also assessed according to the three trimesters of pregnancy, the results of which are presented in Figure 1. The mean body mass index (BMI) of the participants was recorded as 29.45 ± 3.39 kg/m², as shown in Table 2. During the diagnostic process of the study, all patients underwent ultrasound (USG) examination first and then MRI examination for comparative validity. USG gave a positive diagnosis of spina bifida in 82 patients, while MRI confirmed the diagnosis in 83 patients. Of the cases in which USG gave a positive result, 58 were recorded as true positives and 4 as false positives, and of the patients in which USG gave a negative result, 58 were recorded as true negatives and 5 as false negatives, all of which are summarized in Table 3. The diagnostic accuracy of USG was compared with MRI examination results, which were used as the gold standard for postnatal MRI results. The statistical values of USG for the diagnosis of spina bifida were as follows: sensitivity 92.06%, specificity 93.55%, positive predictive value 93.55%, negative predictive value 92.06%, and overall diagnostic accuracy 92.80%. These results indicate that USG is a relatively accurate and reliable technique for the initial diagnosis of spina bifida, but MRI is still considered necessary for confirmation due to its high accuracy.

Table 1: Distribution of patients according to Age (n=125).

Age (years)	No. of Patients	%age
19-30	83	50.40
31-43	82	49.60
Total	165	100.0

Mean ± SD = 30.19 ± 6.53 years

The age distribution of the participants in this table shows that out of the total 165 participants, 83 (50.40%) were between the ages of 19 and 30, and the remaining 82 (49.60%) were between the ages of 31 and 43. This balance shows that cases of spina bifida are seen in both age groups, but little focus has been placed on the condition of young pregnant women. This reflects the fact that this disorder is detectable in the early stages of pregnancy, and therefore the availability of diagnostic facilities for young mothers should be expanded.

Table 2: Distribution of patients according to BMI (n=125).

BMI (kg/m2)	No. of Patients	%age
≤30	77	45.60
>30	88	54.40

Mean ± SD = 29.45 ± 3.39 kg/m2

The BMI table shows that the body mass index of the participants is mostly above 30, where 54.40% of the participants are included in the overweight or obese category. This suggests that obesity may be associated with the incidence of spina bifida, or at least may affect the quality of sonographic diagnosis. Because there are technical limitations in performing accurate ultrasound in patients with high BMI. Therefore, this factor should be seriously considered during diagnosis.

Table 3: Accuracy of Ultrasound for Spina Bifida Detection in First and Second Trimester (vs. Postnatal MRI)

	Positive result on MRI	Negative result on MRI	P-value
Positive on USG	58 (TP)	04 (FP)	0.0001
Negative USG	05 (FN)	58 (TN)	

TP=True positive FP=False positive FN=False negative TN=True negative

This table compares the results of USG with the "gold standard" MRI. The sensitivity (92.06%) and specificity (93.55%) of USG indicate that this technique has a high accuracy for diagnosing spina bifida. The positive and negative predictive values are also reliable, being 93.55% and 92.06%, respectively. This result means that USG, when used by experienced specialists, can be a reliable tool for diagnosis, even in areas where access to MRI is limited. The statistical significance (P-value = 0.0001) indicates that this result is not a coincidence, but rather scientifically valid.

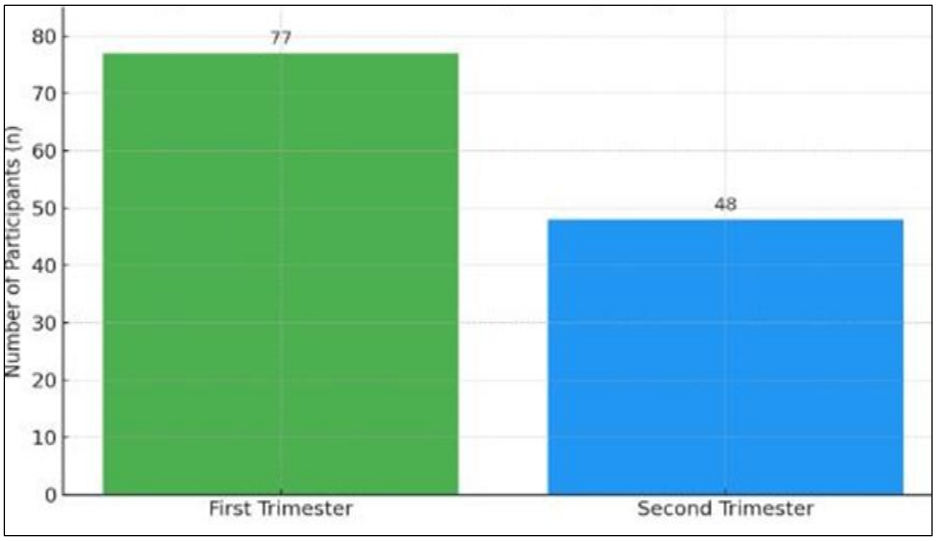

FIG 1: Distribution of Participants by Pregnancy Trimester

Figure 1 shows that 97 out of 165 participants (61.60%) were assessed in the first trimester of pregnancy, and the remaining 68 (38.40%) in the second trimester. This result indicates that more attention is paid to early diagnosis, which is considered the most important stage of prenatal diagnosis for spina bifida. It also suggests that prenatal monitoring and testing should begin in the first weeks of pregnancy, to reduce the need for more advanced and expensive diagnostic steps.

Discussion

Spina bifida, also known as open or closed spina bifida, is a disorder caused by the improper closure of the spinal canal during fetal development. The condition has open (such as myelomeningocele and meningocele) and latent forms, and may present with or without neurological deficits. The open form, which causes serious neurological problems, occurs in 0.5–0.8 per 1,000 live births. Environmental and genetic factors play a role in the development of the condition. Folate deficiency, maternal diabetes, zinc deficiency, alcohol consumption during the first trimester of pregnancy, and use of drugs such as carbamazepine and valproic acid have been implicated as risk factors. This deficiency can be prevented if the mother takes folic acid from three months before conception until the third trimester.

Both prenatal (in utero) and postnatal interventions are available to treat this condition. According to the MOMS study, prenatal surgery significantly improves the mental and physical development of the child. In this study, fetuses with symptoms of spina bifida that appeared between the first and second trimesters were included. Therefore, an accurate assessment of the extent of the lesion is essential for the surgical decision. Ultrasound (USG) is a basic and important tool for the diagnosis of fetal spina bifida, which can detect 80% to 100% of cases. Between the 11th and 14th weeks of pregnancy, measurement of intracranial translucency is considered an effective screening tool. In fetuses with spina bifida, the fourth ventricle is absent, because the development of the hindbrain is abnormal. The second and third trimesters are the best times to examine a direct spinal cord injury, whether one-, two-, or three-dimensional USG is used.

Consultation with the mother, her family, and the professional team is essential, as the location of the injury is important for assessing the patient's likely future position, mobility, and bladder and rectal control. 2D ultrasound can accurately determine the extent of the injury in only 38% of cases after birth, but this can be increased to 96% for two levels and 100% for three levels in skilled centers. However, Bruner and colleagues noted that this method has limitations in determining the exact location of the injury in some cases, and errors of up to three levels are possible, which can affect the decision to intervene, and make it difficult to assess the patient's future mobility, strength, and control abilities. Although 3D ultrasound has been shown to be important for detecting fetal defects, its accuracy compared to 2D is still under debate. In this study, the accuracy of USG assessment was evaluated using postnatal MRI as the gold standard. Ultrasound and subsequent MRI were performed for all patients. USG gave a diagnosis of spina bifida in 62 patients. MRI confirmed the defect in 63 patients. Of the USG-positive cases, 58 were true positives and 4 were false positives. Of the negative cases, 58 were true negatives and 5 were false negatives. Finally, the sensitivity, specificity, positive and negative predictive values, and overall accuracy of ultrasound diagnosis were measured as follows: Sensitivity:

92.80%, Specificity: 92.06%, Positive value: 93.55%, Negative value: 92.06%, Overall accuracy: 92.80%.

Romero *et al*. demonstrated that USG has a sensitivity of 94.7% and a specificity of 98.3%. A fetus with venticulomegaly, omphalocele, severe kyphosis, and scoliosis was misdiagnosed as having spina bifida. Goswami showed that in the interior of Sindh, Pakistan, only 20% of cases of neural tube defects were diagnosed prenatally, and the remaining 80% were diagnosed postnatally. The "banana sign" of the cerebellum and the "lemon sign" of the facial features are indicators of open spina bifida. Closed spina bifida, compared with open, shows fewer changes, so its diagnosis is difficult by ultrasound. 2D USG achieves a sensitivity and specificity of almost 100% for the open type, but this figure decreases for the closed type. In Denmark, 88.5% of SB patients are diagnosed before 22 weeks of gestation, and 93.9% at any stage. According to the EUROCAT report, prenatal diagnosis rates ranged from 81% to 90%, while in this study this figure was 89.3%, indicating the improved quality of the Danish program. After diagnosis, the termination of pregnancy (ToP) rate was also high: 81.6% for all SB cases, 90.3% for those diagnosed before 22 weeks. These figures are higher than EUROCAT (66%), but similar to France (97%) and Italy (92%). In the US state of Georgia, the third trimester diagnosis rate was 34%, and in the north of the Netherlands 78.6%. Recently, with ultrasound, genetic counseling is offered to parents of SB fetuses, although previously this was only done for hereditary diseases. One in ten cases of isolated SB have a chromosomal problem, so all mothers who have a fetus with SB should receive chromosomal testing and advice from a specialist team.

Conclusion

The results of this study clearly demonstrate that ultrasonography is an effective, accurate, and reliable diagnostic technique for prenatal diagnosis of spina bifida. The study is based on data from 165 pregnant women, all of whom underwent USG first and then MRI. The results showed that USG has a high concordance with MRI; the sensitivity of this technique was 92.06%, specificity 93.55%, positive predictive value (PPV) 93.55%, negative predictive value (NPV) 92.06%, and overall diagnostic accuracy 92.80%. These statistical indicators indicate that USG, if performed by experienced specialists, can play a reliable role in the early diagnosis of spina bifida. On the other hand, USG is an economical, non-invasive, and technically more accessible tool, especially in countries and regions where access to MRI is limited or expensive. It should also be noted that a large number of women among the participants were in the first or second trimester of pregnancy, indicating that early diagnosis of spina bifida is possible during these periods. Therefore, this study suggests that ultrasound should be part of "universal screening" for prenatal screening, so that early diagnosis of abnormalities is possible and appropriate medical intervention can be provided. Also, this study highlights the fact that body mass index (BMI) and age structure affect the accuracy of diagnosis, but USG still provided superior results based on its technical reliability. This diagnostic ability further increases the confidence in the ultrasound technique, especially in situations where early diagnosis is vital for treatment, counseling, and family decision-making. The study recommends that USG should be considered a first-line tool for the diagnosis of spina bifida, and health policymakers should promote the widespread use of this technique to ensure prenatal fetal health and improve the quality of life of the mother and child.

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