Original Article

Diagnostic Accuracy of Ultrasonography in Detecting Spinal Dysraphism in Infants, Taking MRI as the Gold Standard

Nosheen Ahmad, Amna Rehan, Abdul Rauf, Mahreen Rasool, Hassan Bukhari, Fizza Kamran

ABSTRACT

Objective: To determine the diagnostic accuracy of ultrasonography in detecting spinal dysraphism in infants, taking MRI as the gold standard.

Methodology: This cross-sectional study was conducted over a period of six months, from 30th May 2023 to 29th November 2023, in the Department of Radiology, Allied Hospital, Faisalabad. A total of 142 infants with suspected spinal dysraphism were included in the study. Newborns with clinically suspected birth-related spinal cord injuries and absolute contraindications to MRI were excluded from the study. Ultrasonography was performed using real-time USG scanners equipped with high-frequency linear array transducers ranging from 9 to 12 megahertz (MHz) for optimal imaging in infants. The USG findings were then compared with MRI results to assess diagnostic accuracy of ultrasonography.

Results: We observed that the overall sensitivity was 91.43%, specificity 94.44%, positive predictive value (PPV) 94.12%, negative predictive value (NPV) 91.89%, and diagnostic accuracy of ultrasonography in detecting spinal dysraphism in infants was 92.96.

Conclusion: Ultrasonography provides a highly sensitive and accurate diagnostic assessment while detecting spinal dysraphism. Being a cost-effective, simple and widely accessible alternative to MRI, it is highly useful modality in low-resource setups.

KEYWORDS: Diagnostic accuracy, Sensitivity, Spinal dysraphism, Ultrasonography.

INTRODUCTION

Spinal dysraphism encompasses a spectrum of congenital spinal anomalies that results from developmental defects during embryogenesis, especially because of disruptions in the complex process of spinal axis formation between the 2nd and 6th weeks of gestation.[1-3] This condition arises from incomplete midline closure of mesenchymal, osseous, and nervous tissue during neurulation, affecting both the primary and secondary stages of neural tube development.

Spinal dysraphism is widely classified into two types: aperta, involving visible lesions, and occulta, which does not present with external abnormalities. As the most prevalent congenital anomaly of the central nervous system (CNS), spinal dysraphism has significant clinical implications, including neurogenic bladder dysfunction.[4,5] Children with spinal dysraphism often face a range of complications, making it a substantial public health concern that not only impacts affected individuals but also places a considerable burden on their families and communities.[6] The

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role of ultrasound in evaluation of spinal dysraphism is pivotal.[7-10]

Research has demonstrated the diagnostic efficacy of spinal ultrasound. One study reported spinal dysraphism in 60.6% of cases and found that the overall accuracy of ultrasonography compared to MRI was high, with specificity ranging from 98.6% to 100%, sensitivity between 66.6% and 91.6%, a positive predictive value (PPV) of 90% to 100%, and a negative predictive value (NPV) of 94.1% to 98.7%.[11] Another study evaluating the accuracy of spinal ultrasound in detecting spinal dysraphism reported specificity between 94.5% and 100%, sensitivity ranging from 81.8% to 100%, PPV between 84.3% and 100%, and NPV between 86.7% and 100%, when compared to MRI.[12]

In poor resource nations like Pakistan where the prevalence of poverty is high and the healthcare and educational facilities are poor and consanguineous marriages are on the rise, the incidence of spinal dysraphism in neonates is high as well.[13,14] Early and accurate diagnosis of spinal dysraphism is highly essential for proper management, so this study was designed to find out the diagnostic accuracy of ultrasonography in detecting spinal dysraphism in infants, using MRI as the gold standard. This study will make a valuable contribution to the literature by providing the local data on the use of ultrasonography. If a high diagnostic accuracy is demonstrated, then based on these findings, ultrasonographic estimation of each suspected infant can be chosen routinely for making proper management decision. Furthermore, it will also assist the clinicians in making proper management plans to help reduce morbidity and mortality in affected infants.

METHODOLOGY

This cross-sectional study was conducted at the Department of Radiology, Allied Hospital, Faisalabad, over a six-month period from May 30, 2023, to November 29, 2023. The study aimed to determine the diagnostic accuracy of ultrasonography (USG) in detecting spinal dysraphism in infants, using magnetic resonance imaging (MRI) as the gold standard. A total of 142 cases were enrolled based on a sample size calculation using the WHO calculator for sensitivity and specificity. The calculation was performed with a 95% confidence level, considering an expected prevalence of spinal dysraphism of 60.6%, a desired sensitivity of 66.6% with a 10% margin of precision, and a specificity of 98.6% with a 3% margin of precision. Patients were selected using a non-probability consecutive sampling technique.

The study included all infants from birth to one year of age who presented with clinical signs suggestive of spinal dysraphism. High-risk indicators included sacral dimples larger than 5 mm in diameter and located more than 2.5 cm above the anus, palpable subcutaneous masses, hemangiomas, areas of abnormal skin pigmentation, skin tags or tails, hairy patches, and sinus tracts. Infants with clinically suspected birth-related spinal cord injuries or those with absolute contraindications to MRI were excluded from the study. Ethical approval was obtained from the institutional review board having reference number 48.ERC/FMU/2023-241543, and informed consent was secured from the parents or legal guardians before enrollment.

All enrolled infants underwent ultrasonography using real-time USG scanners equipped with high-frequency linear array transducers (9–12 MHz) to ensure optimal image resolution. In cases where a broader field of view was necessary or when the acoustic window was restricted, particularly in older infants, a curvilinear probe (3–9 MHz) was utilized. The ultrasound examination was conducted with the infant positioned in the prone position to facilitate better visualization of the spinal structures. When necessary, a modified prone position, such as the reversed Trendelenburg or upright position, was utilized to optimize cerebrospinal fluid (CSF) distension and allow for better visualization of the cauda equina region. Knee flexion was used to enhance separation of the spinous processes for an adequate assessment of the spinal canal contents. A consultant sonologist with at least 5 years of post-fellowship experience performed ultrasonographic examination, using pre-defined criteria to document the presence or absence of spinal dysraphism.

All infants were evaluated through MRI after performing ultrasonographic examination, by using a 1.5 Tesla machine with an 8-channel dedicated phased array coil. The imaging protocol included sagittal and axial sequences of the lumbar and sacral regions, with a field of view (FOV) of 200–250 mm, a matrix size of 265 × 265, slice thickness of 3 mm, and an inter-slice interval of 0.5–1.0 mm. MRI films were also interpreted by a consultant radiologist, and the findings were documented. The results of both modalities were compared following reference standards to know the accuracy of USG.

The collected variables included demographic characteristics such as infant weight in addition to gender and age along with the presence or absence of spinal dysraphism as determined by MRI and ultrasonography (USG). Statistical analysis was performed using SPSS version 25.0 for recording demographics and other findings. We used 2x2 contingency table to record and evaluate diagnostic performance of ultrasonography, measuring sensitivity, specificity, positive predictive value, negative predictive value and overall diagnostic accuracy with MRI serving as the gold standard.

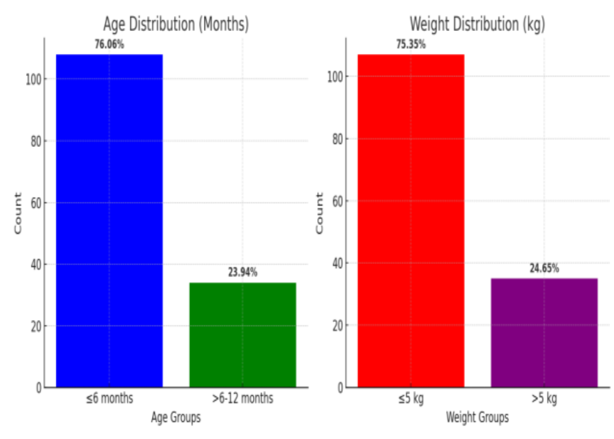
RESULTS

The age range in this study was from birth to 12 months, with a mean age of 5.05 ± 2.23 months. The majority of the patients (76.06%) were ≤6 months old, while the remaining 23.94% were aged between 6 and 12 months. The distribution of patients according to gender is illustrated in the corresponding figure. The mean weight of the infants was 4.63 ± 1.29 kg. (Table & Fig. 1) The overall diagnostic accuracy of ultrasonography in detecting spinal dysraphism, using MRI as the gold standard, was found to be 92.96%. The sensitivity of ultrasonography was 91.43%, indicating its ability to correctly identify infants with spinal dysraphism, while the specificity was 94.44%, reflecting its ability to correctly rule out those without the condition. The positive predictive value (PPV), representing the proportion of true positive cases among all positive results, was 94.12%, and the negative predictive value (NPV), representing the proportion of true negative cases among all negative results, was 91.89%.

Table I: Distribution of Patients by Age and Weight (n=142)

Variable		Count	Percentage
Age(months)	≤6	108	76.06%
	>6-12	34	23.94%
Total		142	100%
Weight (kg)	≤5	107	75.35%
	>5	35	24.65%
Total		142	100%

When stratified by age, the sensitivity and specificity of ultrasonography were slightly higher in infants ≤6 months (91.67% and 95.00%, respectively) compared to those >6–12 months (90.91% and 91.67%, respectively). The PPV and NPV were also higher in the younger age group (93.62% and 93.44%) compared to the older group (95.24% and 84.62%).



Gender-based stratification revealed that diagnostic accuracy was slightly higher in males (92.42%) compared to females (91.53%). Sensitivity was 90.91% in males and 87.10% in females, while specificity was 93.18% and 96.43%, respectively. The PPV was higher in females (96.43%) compared to males (86.96%), whereas the NPV was higher in males (95.35%) compared to females (87.10%).

Table II: Diagnostic Accuracy of Ultrasonography						
Diagnostic accuracy		Sensitivity	Specificity	PPV	NPV	Diagnostic Accuracy
Overall		91.43 %	94.44 %	94.12 %	91.89 %	92.96%
Age (months)	≤6	91.67 %	95.00 %	93.62 %	93.44 %	93.52%
	>6-12	90.91 %	91.67 %	95.24 %	84.62 %	91.18%
Gender	Male	90.91 %	93.18 %	86.96 %	95.35 %	92.42%
	Female	87.10 %	96.43 %	96.43 %	87.10 %	91.53%
Weight (kgs)	≤5	90.20 %	94.64 %	93.88 %	91.38 %	92.52%
	>5	94.74 %	93.75%	94.74 %	93.75 %	94.29%

Weight-based stratification showed that ultrasonography had a slightly higher diagnostic accuracy in infants weighing more than 5 kg (94.29%) compared to those weighing 5 kg or less (92.52%). The sensitivity, specificity, PPV, and NPV were also higher in the heavier weight group (94.74%, 93.75%, 94.74%, and 93.75%, respectively) compared to those in the lighter group (90.20%, 94.64%, 93.88%, and 91.38%, respectively). (Table 2)

These findings indicate that ultrasonography demonstrates high diagnostic accuracy in detecting spinal dysraphism across different age groups, genders, and weight categories, with minor variations in sensitivity and specificity. The results support its potential use as an effective screening tool for early detection of spinal dysraphism in infants.

DISCUSSION

MRI is recognized widely as the most informative imaging modality for assessing spinal dysraphism, as it provides a detailed view of spinal cord anatomy. It offers superior visualization of the interface between lipomas and the spinal cord compared to CT myelography. Sagittal MRIs have been found to be more effective than CT myelograms in delineating these structures. However, MRI does not optimally visualize individual nerve roots.[15-17] Furthermore, its limited availability in low resource setup and associated cost restricts its benefits.

In neonatal assessment, ultrasonography plays a crucial role, particularly in the initial evaluation of spinal defects.[18-20] Although its application for assessing the spinal canal is primarily limited to the neonatal period, it can still provide clear imaging of soft tissue-covered defects even in adulthood. Increasingly, fetal ultrasonography is being used as the primary screening tool for neural tube defects (NTDs), typically around the 18th week of gestation. This shift reflects growing confidence in ultrasonography's accuracy and safety, particularly since it eliminates the 1% miscarriage risk associated with invasive procedure, such as amniocentesis.[21]

Our study demonstrated an overall diagnostic accuracy of 92.96% for ultrasonography in detecting spinal dysraphism, using MRI as the gold standard. The sensitivity was 91.43%, and the specificity was 94.44%, with a PPV of 94.12% and an NPV of 91.89%. Various prior studies are aligned with our findings regarding accurate diagnosis of spinal ultrasound.[11,12]

A previous study in 2020 documented an excellent agreement between MRI and ultrasound in cases involving infants aged ≤ 2 years, by calculating higher sensitivity range i.e., 84.3% to 100% while the specificity ranged between 94.5% and 100%. In our study, the calculated sensitivity of 91.43% falls within this range, indicating that ultrasonography (USG) demonstrates high diagnostic accuracy in detecting spinal dysraphism in infants. Moreover, our specificity of 94.44% is in agreement with Tawfik and others 'findings, reinforcing the hypothesis that ultrasound can effectively diagnose spinal dysraphism in most cases.[12]

Similarly, Keykhosravi et al. (2023), in a study aimed at explaining the role of ultrasonography in the diagnosis of tethered cord syndrome (TCS) — a form of occult spinal dysraphism — highlighted its effectiveness as a valuable diagnostic tool.[22] Their findings reveal that ultrasonography effectively distinguished TCS patients from healthy individuals, highlighting remarkable differences in spinal cord positioning. This suggests that ultrasonography can be particularly useful for detecting subtle spinal abnormalities early.

Our study also evaluated the diagnostic accuracy of ultrasonography across different age groups, genders, and weight categories:

- Sensitivity and specificity were higher in infants ≤6 months (91.67% and 95.00%) compared to those >6–12 months (90.91% and 91.67%).
- Diagnostic accuracy was slightly higher in males (92.42%) compared to females (91.53%).
- Accuracy was higher in infants weighing more than 5 kg (94.29%) compared to those weighing ≤5 kg (92.52%).

These variations in diagnostic accuracy are consistent with the findings of Tawfik et al. (2020), who observed that ultrasound accuracy declines as infants grow older due to increased vertebral ossification.[12] This supports the recommendation that ultrasonography should ideally be performed in the first few months of life, before significant spinal ossification occurs.

Recent technological advancements in imaging, such as 3D ultrasound and AI-driven algorithms, have enhanced the accuracy of ultrasonography. Charis et al. (2025) discussed how AI-assisted ultrasonography has improved the detection rates of spinal dysraphisms, particularly in cases where fetal positioning poses challenges.[8] These advancements indicate a promising future for ultrasound in screening for spinal dysraphism, potentially reducing the dependency on MRI in certain cases. However, MRI remains the gold standard for evaluating spinal dysraphism, particularly for detailed soft tissue assessment and surgical planning. Tawfik et al. (2020) emphasized that MRI is crucial for confirming ultrasonography findings and detecting additional spinal anomalies that may influence surgical decisions.[12]

Recent literature continues to support the efficacy of ultrasonography for spinal dysraphism diagnosis. Gajagowni et al. (2024) evaluated the diagnostic yield of spinal ultrasound in neonates with sacral dimples.[23] Their findings revealed that spinal ultrasound had a low diagnostic yield when performed for sacral dimples alone but remained a valuable tool for detecting caudal regression syndrome and other complex malformations. This suggests that while ultrasonography is useful, its diagnostic performance may be influenced by the specific indications for screening.

Komatsu et al. (2021) explored the application of artificial intelligence (AI) in fetal ultrasound analysis.[24] Their study demonstrated that deep learning algorithms improved the accuracy of detecting fetal structural abnormalities, with AI-assisted analysis showing higher specificity compared to conventional ultrasound methods. This highlights the potential for integrating AI-based enhancements in spinal ultrasound to further improve diagnostic accuracy.

Another study by Shahrivar co-workers in the year 2023 performed a comprehensive review on machine learning applications in ultrasonography.[25] They are of the view that advanced image technology could significantly improve anomaly detection in prenatal and neonatal imaging. These findings advocate that future improvements in spinal ultrasound technology may include AI-assisted anomaly detection, which may further improve specificity as well as sensitivity.

CONCLUSION

Ultrasonography, being a cost-effective, non-invasive, and readily accessible imaging modality, demonstrated high diagnostic accuracy in detecting spinal dysraphism in infants.

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Nosheen Ahmad: conceived the study designed, carried out the data collection and statistical analysis and drafted the manuscripts.

Amna Rehan: Participated in its design and coordination. drafted, read and approved the final manuscript.

Abdul Rauf: Participated in its design and coordination. Statistical analysis, drafted, read and approved the final manuscript.

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