



Diagnostic Accuracy of Ultrasound in Diagnosing Spinal Dysraphism in Children Keeping Magnetic Resonance Imaging as Gold Standard

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Authors' Contribution

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ABSTRACT

Background: Spinal dysraphism is a congenital condition when the spine and spinal cord do not develop normally which potentially leading to severe complication if not detected early. Ultrasound is a common diagnostic tool in infant but its accuracy is still debated with MRI considered the gold standard. **Objective:** To assess the diagnostic accuracy of ultrasound in detecting spinal dysraphism in children taking MRI as the gold standard. **Study Design:** Cross-sectional study. **Duration and Place of Study:** This study was conducted from November 2023 to May 2024 at Liaquat University of Medical and Health Sciences, Pakistan. **Methodology:** A total of 253 children aged 1 month to 2 years with suspected spinal dysraphism were enrolled. Ultrasound was performed to detect low-lying conus medullaris, thickened filum terminale and other markers. MRI was used as the reference standard. The diagnostic parameters sensitivity, specificity, positive predictive value and negative predictive value were calculated. **Results:** The ultrasound showed sensitivity of 87.30%, specificity of 71.70% and a diagnostic accuracy of 75.10%. The PPV was 46.20% and the NPV was 95.30%. Stratified results showed higher sensitivity in younger children (88.00%) and males (95.00%). **Conclusion:** Ultrasound is an effective screening tool for spinal dysraphism especially in excluding the condition due to its high NPV.

INTRODUCTION

Spinal dysraphism is a group of congenital defect where the spine and spinal cord do not form in a normal way during early pregnancy.¹ It includes open and closed forms such as spina bifida aperta, meningocele, myelomeningocele and occult spinal dysraphism with skin sign.² In many children the defect can be hidden under the skin and it may present later with problems like weak legs, abnormal gait, back swelling, foot deformity, bowel or bladder issues or repeated urinary infection.³ Some infants show midline skin markers over the lower back like a deep dimple, tuft of hair, hemangioma, lipoma, skin tag or deviated gluteal cleft and these signs can raise suspicion for an underlying spinal cord problem.⁴ If spinal dysraphism is not recognized early tethered cord and related nerve damage can progress with growth so early detection is important for timely follow up and surgical planning when needed.⁵

For diagnosis magnetic resonance imaging is considered the gold standard because it show the spinal cord, nerve roots, conus level, filum terminale and associated lesions in very good detail.⁶ MRI can detect tethered cord, thickened filum, lipomyelomeningocele,

dermal sinus tract, split cord malformation, syrinx and other changes and it also helps to map the anatomy before operation.⁷ The diagnostic accuracy of any test is judged by comparing it with MRI results using measures like sensitivity, specificity, positive predictive value and negative predictive value and also by looking at false negative and false positive findings.⁸

Ultrasound is commonly used as an early imaging tools for suspected spinal dysraphism in infants mainly before the posterior elements of the spine fully ossify.⁹ In young babies spinal ultrasound can show the level of conus medullaris, movement of the cord, thickness of filum terminale, presence of cystic mass, lipoma like echogenic tissue and indirect signs of tethering so it can act as a screening method.¹⁰ When MRI is kept as gold standard ultrasound may show good sensitivity for some findings like low lying conus and obvious cystic lesions but it can miss small dermal sinus tracts, subtle split cord or deep complex anomalies and this reduce its diagnostic accuracy in some cases.¹¹ The accuracy also depend on the operator skill, machine quality and child age because after a few months the bony spine reduce the window and the images become less clear.¹² According to a study, spinal

dysraphism was found in 23% of the cases.¹³ Ultrasound (US) has sensitivity of 86% and specificity of 55% in diagnosing spinal dysraphism.¹⁴

There is a need to do a study on the diagnostic accuracy of ultrasound in diagnosing spinal dysraphism in children because this condition can lead to serious complications if not detected early. Many children may have undiagnosed spinal dysraphism, and using ultrasound as a diagnostic tool could help in identifying the condition quickly and accurately. A study would provide better understanding of how reliable ultrasound is in detecting this issue in children and how it can improve early diagnosis and treatment outcomes.

METHODOLOGY

This study was done at the Department of Radiology at Liaquat University of Medical and Health Sciences from 1st November 2023 to 1st May 2024. The sample size was calculated using Dr. Lin Naing's sensitivity and specificity calculator. The expected sensitivity was considered as 86%, specificity as 55%,¹⁴ and spinal dysraphism frequency as 23%,¹³ with a 95% confidence interval and 9% margin of error. The sample size for sensitivity was 253 and for specificity was 154, so the larger sample size of 253 was chosen. The study design was a cross-sectional and non-probability consecutive sampling method was used. The patients included were male and female aged between 1 month and 2 years showing signs of spinal dysraphism like weakness in any limb or pain in back or leg for two weeks or more. The exclusion criteria were patients already diagnosed with spinal dysraphism or who had MRI done first and then ultrasound. Patients with central nervous system infections were also excluded. Before starting, ethical approval was obtained from the Institutional Review Board (No. LUMHS/REC/-107, Dated 26-07-2023) and the College of Physicians and Surgeons of Pakistan. Informed consent was taken from all participants. Data was collected through a questionnaire, and patients were enrolled from the ultrasound department of LUMHS. Demographics and clinical details including antenatal folic acid supplementation history were recorded. Ultrasound was performed by a fellowship-trained radiologist with over 5 years of experience. The MRI was done on a 1.5 Tesla machine with sequences including axial T1, axial T2, coronal FLAIR and post-contrast T1 images. MRI was reported by a radiologist with more than 5 years of post-fellowship experience. For diagnosing spinal dysraphism ultrasound was considered positive if any of the following were observed: low-lying conus medullaris (at or below L2 level), lipoma or fibrolipoma of the filum terminale, thickened filum terminale or presence of meningocele, myelocele or myelomeningocele. If these same findings were seen on MRI it was also considered positive.

Data analysis was done using SPSS 21. Quantitative data such as age and symptom duration were expressed as mean \pm standard deviation. Qualitative data like gender, place of residence (urban or rural), antenatal folic acid intake and spinal dysraphism findings on ultrasound and MRI were presented as frequency and percentages. The diagnostic accuracy, sensitivity, specificity, positive predictive value and negative predictive value of

ultrasound in diagnosing spinal dysraphism were calculated using a 2x2 table with MRI as the gold standard. Stratification was done based on effect modifiers like age, gender, symptom duration to see how they affected the outcomes. Post-stratification the sensitivity, specificity, PPV, NPV and diagnostic accuracy were recalculated.

RESULTS

The study enrolled 253 patients with mean age of 13.51 ± 6.98 months and mean duration of symptoms being 37.48 ± 14.04 days (as shown in Table-I). The gender distribution was showing that 76 patients (30.0%) were males while 177 patients (70.0%) were females. Regarding place of residence, 137 patients (54.2%) belonged to urban areas and 116 patients (45.8%) were from rural areas. The antenatal folic acid intake was reported in 126 patients (49.8%) whereas 127 patients (50.2%) had no history of folic acid intake during pregnancy (as shown in Table-I).

Table I
Patient Demographics

Demographics	Mean \pm SD
Age (months)	13.51 \pm 6.98
Duration of Symptoms (days)	37.48 \pm 14.04
Gender	
Male n (%)	76 (30.0%)
Female n (%)	177 (70.0%)
Place of Residence	
Urban n (%)	137 (54.2%)
Rural n (%)	116 (45.8%)
Antenatal Folic Acid Intake	
Yes n (%)	126 (49.8%)
No n (%)	127 (50.2%)

The overall results was demonstrating that ultrasound detected spinal dysraphism in 61 patients (24.1%) while it was negative in 192 patients (75.9%). On the other hand, MRI which was taken as gold standard showed positive findings in 55 patients (21.7%) and negative findings in 198 patients (78.3%) out of total 253 patients (as shown in Table-II).

Table II
Overall results of Ultrasound and MRI in diagnosis of spinal dysraphism

Spinal Dysraphism	Ultrasound	MRI
Positive	61 (24.1%)	55 (21.7%)
Negative	192 (75.9%)	198 (78.3%)
Total	253 (100%)	253 (100%)

When ultrasound was compared with MRI, the results revealed that out of 104 positive ultrasound cases, 48 were true positive and 56 were false positive. Among 149 negative ultrasound cases, 7 were false negative and 142 were true negative. The total MRI positive cases were 55 while MRI negative cases were 198 (as shown in Table-III).

Table III
Comparison of Ultrasound versus MRI in diagnosis of spinal dysraphism

Ultrasound	MRI		Total
	Positive	Negative	
Positive	48 (TP)	56 (FP)	104
Negative	7 (FN)	142 (TN)	149
Total	55	198	253

Key: TP = True positive, FP = False positive, FN = False negative, TN = True negative

The diagnostic parameters of ultrasound was showing sensitivity of 87.30%, specificity of 71.70%, and diagnostic accuracy of 75.10%. The positive predictive value was 46.20% while negative predictive value was 95.30% (as shown in Table-IV).

Table IV

Sensitivity, Specificity, Diagnostic Accuracy, PPV and NPV of Ultrasound in diagnosis of spinal dysraphism

Diagnostic Parameter	Result
Sensitivity	87.30%
Specificity	71.70%
Diagnostic Accuracy	75.10%
PPV	46.20%
NPV	95.30%

In stratified analysis according to age, for patients aged ≤ 12 months, the sensitivity was 88.00%, specificity was 77.30%, diagnostic accuracy was 80.20%, PPV was 59.50% and NPV was 94.40%. For patients aged >12 months, sensitivity was 86.70%, specificity was 68.90%, diagnostic accuracy was 72.20%, PPV was 38.80% and NPV was 95.80%. When stratified by gender, male patients showed sensitivity of 95.00%, specificity of 73.20%, diagnostic accuracy of 78.90%, PPV of 55.90% and NPV of 97.60%. Female patients demonstrated sensitivity of 82.90%, specificity of 71.10%, diagnostic accuracy of 73.40%, PPV of 41.40% and NPV of 94.40%. According to duration of symptoms, for ≤ 28 weeks duration, sensitivity was 88.00%, specificity was 75.00%, diagnostic accuracy was 78.80%, PPV was 59.50% and NPV was 93.80%. For >28 weeks duration, sensitivity was 86.70%, specificity was 70.30%, diagnostic accuracy was 73.20%, PPV was 38.80% and NPV was 96.00% (as shown in Table-V).

Table V

Stratified analysis of Sensitivity, Specificity, Diagnostic Accuracy, PPV and NPV of Ultrasound in diagnosis of spinal dysraphism with age, gender and duration of symptoms

Variables	Groups	Diagnostic Parameter	Result
Age (months)	≤ 12	Sen	88.00%
		Spec	77.30%
		DA	80.20%
		PPV	59.50%
		NPV	94.40%
	>12	Sen	86.70%
		Spec	68.90%
		DA	72.20%
		PPV	38.80%
		NPV	95.80%
Gender	Male	Sen	95.00%
		Spec	73.20%
		DA	78.90%
		PPV	55.90%
		NPV	97.60%
	Female	Sen	82.90%
		Spec	71.10%
		DA	73.40%
		PPV	41.40%
		NPV	94.40%
Duration of Symptoms (weeks)	≤ 28	Sen	88.00%
		Spec	75.00%
		DA	78.80%
		PPV	59.50%
		NPV	93.80%
	>28	Sen	86.70%
		Spec	70.30%
		DA	73.20%
		PPV	38.80%
		NPV	96.00%

DISCUSSION

The present study was found that ultrasound has sensitivity of 87.30%, specificity of 71.70% and diagnostic accuracy of 75.10% in detecting spinal dysraphism. These findings was suggesting that ultrasound is good screening tool because of high sensitivity which means it can identify most of the positive cases correctly. However, the specificity was relatively lower which indicates that ultrasound may produce false positive results in significant number of cases. The high negative predictive value of 95.30% was demonstrating that when ultrasound shows negative result, it is highly reliable to rule out the disease. The study was enrolled 253 patients with female predominance 177 (70.0%) compared to males 76 (30.0%). This gender distribution was consistent with known epidemiological pattern where spinal dysraphism occurs more frequently in female children due to hormonal and genetic factors that influences neural tube development. The mean age of patients was 13.51 ± 6.98 months which reflects that most cases presents in infancy period when clinical signs becomes more apparent. Interestingly, antenatal folic acid intake was reported in only 126 patients (49.8%) while 127 patients (50.2%) had no history of folic acid supplementation. This was important finding because folic acid deficiency during pregnancy is well established risk factor for neural tube defects including spinal dysraphism. The lack of folic acid intake in about half of the mothers may explains the occurrence of disease in these children.

The current study was found sensitivity of 87.30%, specificity of 71.70% and diagnostic accuracy of 75.10% for ultrasound in diagnosing spinal dysraphism which was comparable with findings of Soliman *et al.*¹⁵ who reported accuracy of $>95.56\%$ in infants with mean age 23.44 ± 32.02 days. However, Soliman *et al.*¹⁵ achieved higher accuracy possibly because their study was included only neonates and infants within first 60 days of life when acoustic window is more favorable, whereas present study was enrolled children with mean age of 13.51 ± 6.98 months where posterior element ossification may have started reducing ultrasound penetration. Similarly, Reda *et al.*¹⁶ also demonstrated high sensitivity ranging from 50% to 100% for different spinal anomalies in neonates and infants aged 1-6 months, which supports the concept that ultrasound performance decreases with increasing age.

The present study was showed that ultrasound detected spinal dysraphism in 61 patients (24.1%) while MRI confirmed it in 55 patients (21.7%), with 48 true positive, 56 false positive and 7 false negative cases. These findings was partially concordant with Dhingani *et al.*¹⁷ who reported that ultrasound and MRI agreed fully in 79.31% cases and partially in 20.69% cases, with MRI-operative concordance being 100% versus 91.30% for ultrasound. The false positive rate in current study was relatively high which explains the lower specificity of 71.70%, and this could be attributed to interpretation errors or artifacts in ultrasound imaging that mimics pathological findings. Tawfik *et al.*¹⁸ stratified their patients by age and found excellent agreement between ultrasound and MRI in children ≤ 2 years with sensitivity 84.3-100% and specificity 94.5-100%, but in children >2

years the agreement was fair and sensitivity dropped to 33.3%. This was supporting the findings of current study where stratified analysis showed that sensitivity was higher in younger age group ≤ 12 months (88.00%) compared to >12 months (86.70%), though the difference was not very marked. The reason for age-related decline in ultrasound accuracy is progressive ossification of posterior spinal elements which creates acoustic shadowing and limits visualization of deeper structures.

The female predominance in current study with 177 patients (70.0%) was consistent with literature where Dhingani *et al.*¹⁷ reported 60.53% females, Reda *et al.*¹⁶ found 53.4% females, Sultana *et al.*¹⁹ documented 64% females, Soliman *et al.*¹⁵ showed 68.9% females and Tawfik *et al.*¹⁸ reported 57.8% females. This gender distribution reflects the established epidemiological pattern of spinal dysraphism where female children are more commonly affected due to hormonal influences on neural tube closure during embryonic development. The antenatal folic acid intake was reported in only 126 patients (49.8%) in present study which highlights the importance of periconceptual folic acid supplementation. This finding was clinically significant because folic acid deficiency is major preventable risk factor for neural tube defects, and inadequate intake in approximately half of the mothers may have contributed to disease occurrence in their children. The positive predictive value in current study was 46.20% which indicates that less than half of positive ultrasound results were confirmed by MRI, whereas negative predictive value was 95.30% showing high reliability of negative ultrasound results in ruling out disease. This pattern

suggests that ultrasound is better screening tool for exclusion rather than confirmation of spinal dysraphism, and positive ultrasound findings should be confirmed with MRI before surgical planning, which is consistent with recommendations of Dhingani *et al.*¹⁷ Reda *et al.*¹⁶ and Soliman *et al.*¹⁵ who all emphasized MRI for pre-surgical mapping despite good screening performance of ultrasound.

The present study was conducted at single center which may limit the generalizability of findings to other populations and settings. The sample size of 253 patients was relatively small and larger multicenter studies are needed to validate these results. The study was not included surgical correlation in all cases which could have provided more accurate assessment of diagnostic parameters. Moreover the interobserver variability between different radiologists was not evaluated which may affect the reproducibility of ultrasound findings. The study was performed in children with mean age of 13.51 months and results may not be applicable to older children where ossification is more advanced.

CONCLUSION

The present study has concluded that ultrasound is effective screening modality for diagnosis of spinal dysraphism in children with good sensitivity and high negative predictive value making it reliable tool for excluding disease. But the lower specificity and positive predictive value indicates that positive ultrasound findings should be confirmed with MRI before any surgical intervention.

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