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Systematic review of ultrasound and MRI prediction of spinal cord anomalies in children with anorectal malformations: what a pediatric urologist writing a protocol needs to know

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ABSTRACT

Background Anorectal malformation (ARM) have a high association with spinal cord anomaly (SCA) impacting bladder and bowel function. This study aims to report the diagnostic accuracy of ultrasound (US) and MRI to detect SCA in children with ARM.

Method A systematic review was performed as per PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidance. Search terms used were broadened in three consecutive searches to find papers investigating outcomes from spinal imaging in ARM, with four author search validation. Study quality was assessed as per Quadas 2 score. Meta-analysis comparing US diagnosis to MRI findings was performed using random effects model, including only clinically relevant SCA and considering children who did not develop negative outcomes as a true negative.

Results Eight studies were included, six reporting US outcomes, and two MRI. All studies but one were retrospective studies. Overall, the included studies were applicable, but all suffered risk of bias with incomplete and non-protocolized follow-up. Pooled analysis for MRI compared with surgery found a sensitivity of 97% and a specificity of 94%, with one false positive and one false negative in an 8-month-old baby. When clinically relevant diagnoses were extracted and meta-analysis performed, a sensitivity of 33% and a specificity of 87% were found for infant US diagnosis of SCA.

Conclusion The normal US spine in early infancy does not have diagnostic weight. A positive early scan will allow you to stream into active surveillance. Mandated MRI of infants will depend on the local approach to prophylactic de-tethering surgery.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Children with anorectal malformation (ARM) are at high risk for spinal dysraphism; common imaging modalities often include ultrasound (US) and MRI.

WHAT THIS STUDY ADDS

⇒ This review highlights that while US can be useful for screening in early infancy, MRI should be used where definitive diagnosis is required.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Depending on local preference, MRI should be considered as part of the screening pathway in children with ARM.

INTRODUCTION

While much of the care of infants with anorectal malformation (ARM) is directed by the pediatric surgeons, the risk of death and morbidity comes from the associations from allied specialities, described by the VACTERL (Vertebral defects, anal atresia, cardiac defects, tracheo-oesophageal fistula, renal anomalies and limb abnormalities) association. The most hazardous being cardiac anomalies, airway collapse and renal failure. Death from renal impairment is estimated at 2.5%–6.0% within the ARM population. Neuropathic bladder is a major risk factor for urinary tract infection and progression toward renal impairment.

Neurogenic bladder is seen in up to 20%–36% of children with an ARM.[1] Neurogenic bladders in the ARM population may be innate, secondary to a spinal defect or a result of nerve damage during corrective PSARP surgery (posterior sagittal anorectoplasty).[2 3] All centers specializing in the care of infants and children with ARM are cognizant of the risk of bladder failure and renal impairment, but protocols for surveillance and management vary.[4 5] In particular, debate remains as to whether plain X-ray for spinal

skeletal anomalies and ultrasound (US) for intraspinal lesions is sufficient to exclude occult spinal anomalies in children with ARM.

This study aims to investigate the efficacy of US and MRI for the diagnosis of spinal anomalies in a systematic fashion and help guide protocol development to aid with the early identification of spinal cord anomaly (SCA) in children with ARM.

METHODS

A systematic review and meta-analysis of diagnostic accuracy was performed according to Cochrane principles[6] and PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidance [7]to investigate the diagnostic accuracy of the imaging modalities for spinal dysraphism for infants with anorectal malformations.

Study selection and data collection

The search strategy was directed as per registered protocol (CRD42021228559). The search was performed using the MeSH (medical subject headings) terms: (anorectal malformation, imperforate anus) AND (neural tube defects) by authors IZ, CM, AE and GK.

The identified papers were filtered by title, abstract and then full papers. Exclusions were made as per standard systematic review methodology.[8] Duplicates were removed, and papers not pertaining to the diagnostic methods, diagnostic accuracy, and the wrong population were removed. Those papers in press were included. Furthermore, papers were assessed for whether diagnostic accuracy was quoted or could be inferred from the reported results; if not, these were excluded. The papers included were decided on after two meetings with the four main authors. No automation tools were used.

Data summary and analysis

The papers were scrutinized for sensitivity and specificity for US or MRI for SCA on a per-patient basis. Data were extracted by hand. Demographics of age, level of ARM ideally based on the Krichenbeck classification,[9] technology details and professional expertise, and associated VACTERL conditions were described. The Quadas2 score[10] was applied and reported for each rating to the included papers to assess risk of bias. Heterogeneity between the datasets was tested with I^2.

A comparison was made of the index test against the reference outcome for US diagnostic accuracy to MRI findings and MRI findings to surgery. A meta-analysis was performed with an interpretation of clinical relevancy, whereby the authors considered tethering, lipoma and filum terminale thickening >2 mm relevant and other findings such as syrinx not clinically relevant. We analyzed the data taking of the authors at face value, accepting non-development of clinical neurological deterioration on mid- and long-term follow-up to represent a "negative outcome." Meta-analysis was performed using Medcalc and Review Manager (RevMan) V.5.4. A random effects model was chosen.

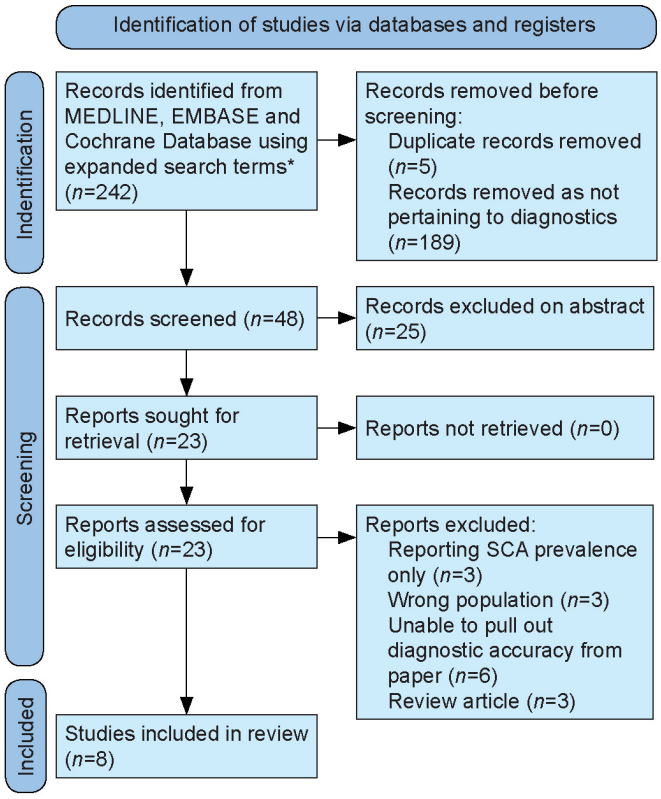

Figure 1 [7]Flow diagram of selection study selection. SCA, spinal cord anomaly.

RESULTS

A total of 242 initial papers were found on the broader MESH term search, and 189 papers were excluded as having no value for inclusion in a systematic review of imaging diagnostics. After further exclusions, as shown in figure 1, 23 full texts were read in full, and eight papers were included in this study.

Description of papers included

Three papers were designed to investigate our specific research question,[11–13] while in five, the data could be inferred.[14–18] These papers described the diagnosis and management of SCA in ARM. Full description of the papers included can be found in table 1. The papers varied in the reporting of SCA from those that reported any anomaly to those that reported a narrow and defined band of 'clinically relevant' SCA. In six papers, US was compared with MRI findings as the reference, in two papers MRI findings were compared with surgery as the reference, and in one, plain radiographs and US were compared with MRI. The significance of the findings was considered differently by the different authors, with four reporting any spinal anomaly as a positive outcome,[11–14] one considering predefined anomalies as a positive outcome,[15] and one considering symptoms along with MRI findings as the defining outcome.[16]

Table 1 Full description of the included papers

Study	Study design	Which imaging was tested? Against which standard?	Definition of spinal cord anomaly	Number undergoing each modality	Number screeningvs responsive to symptoms	Sensitivity	Specificity
Beek *et al.* 1995[13]	Prospective study	Radiographs and spinal US against MRI	Tethered cord, hydrosyringomyelia, lipoma, thick filum terminale	21 X-ray 21 US 21 MRI	All screening Number of symptomatic not specified 16 showed intraspinal anomalies on both US and MRI	100.0%	100.0%
Long *et al.* 1996[19]	Retrospective chart review	MRI to surgery	MMC, tethering, lipoma and diffusely thickened filum (>2 mm)	86 MRI 24 surgeries	62 screening	95.8%	100.0%
Uchida *et al.* 2007[20]	Retrospective chart review	MRI to surgery	Tethered cord	28 MRI 28 surgeries	25 asymptomatic	92.3%	100.0%
Teo *et al.* 2012[18]	Retrospective study	US to MRI	Low-lying cord	101 US 20 MRI	101 screening	70.6%	96.2%
Scottoni *et al.* 2014[14]	Retrospective study	US to MRI	Tethered cord, lipoma, syringomyelia	82 US 82 MRI	All screening	14.8%	100.0%
van den Hondel *et al.* 2016[17]	Retrospective case study	US to MRI	Tethered cord, lipomeningocele, lipoma, meningo (myelo) cele, diastematomyelia, thickened or fatty filum	94 US 17 MRI	94 screened	80.0%	89.0%
Jehangir *et al.* 2020[15]	Retrospective study	US to MRI	Tethered cord, evidence of spinal dysraphism	137 US 20 MRI	All screening	91.0%	75.0%
Esposito *et al.* 2021[16]	Retrospective chart review	US to MRI	MMC, syrinx, arachnoid cyst, conus anomalies, tethered cord	222 US 191 MRI	All screening Number of symptomatic not specified	30.0%	100.0%

MMC, Myelomeningocele; NS, not specified; ref., reference; US, ultrasound.

Table 2 Describing the risk of bias and applicability concerns of the included study as per the QUADAS 2 protocol

Study	Risk of bias				Concerns of applicability		
	Patient selection	Index test	Reference standard	Flow and timing	Patient selection	Index test	Reference standard
Beek *et al.* 1995[13]	Low	Low	Low	Low	Low	Low	Low
Long *et al.*1996[19]	Low	Low	Low	High	Low	Low	Low
Uchida *et al.* 2007[20]	Low	Unclear	Low	Low	Low	Low	Low
Teo *et al.* 2012[18]	Low	Low	Low	Unclear	Low	Low	Low
Scottoni *et al.* 2014[14]	Low	High	Low	Low	Low	Low	Low
van den Hondel *et al.* 2016[17]	High	Low	High	High	Low	Low	Low
Jehangir 2020[15]	Low	Low	High	Unclear	Low	Low	Low
Esposito *et al.* 2021[16]	Low	Low	Low	Unclear	Low	Low	Low

All studies, but one, were retrospective in nature and used single hospital datasets. Overall, the papers were of low risk of non-applicability for our study question (see table 2). Poorly defined reference standards, insufficiently detailed reporting of diagnostic accuracy of those with symptoms and those without and poor length of follow-up lead to concerns of bias trending toward overstating sensitivities in these papers.

Age

The mean age for US was reported as "neonatal" or early in all papers with no exact descriptions of age at the time of scan. The mean age for MRI was given in two papers (2 years 9 months[17] and 3 years 5 months).[18] In these two papers, the differentiation of age of MRI by screening versus responsive to symptoms was not given. Esposito *et al.*[14] screened their patients with MRI at 1 year of age. In all but one study,[18] the duration of follow-up was not given.

Level of anorectal malformation

The Krickenbeck classification was used to describe the level of ARM in two included papers[12 13]; otherwise, the level was described by the Wingspread classification as high, intermediate and low. The distribution of ARM levels within the papers represented the expected rates for the population. One paper stated no association of rates of SCA by level of ARM.[18] Two reported a positive association; as the level became more severe, the likelihood of a spinal anomaly increased, with 44%–57% in high, 36%–41% in intermediate and 17%–27% in low.[14 17]

VACTERL associations

Associations were described in three papers with cardiac, genitourinary and chromosomal anomalies described as expected. No paper correlated spinal findings with vesicoureteric reflux or renal dysplasia. Considering bony anomalies, 13 out of 28 with partial sacral agenesis had tethering,[17] and the sacral ratio on plain film was a good predictor of spinal anomaly. For those with vertebral or sacral anomalies, there was over a 70% chance of some spinal anomaly.[14]

Technology and expertise reviewing imaging

The technology of the MRI scans used was outlined in five papers, and they all adopted T1 and T2 weighted images with slice thickness varying from 3 to 6 mm.[11 14 16 17] The spinal US scans were performed by pediatric sonographers and radiologists. The MRI was reviewed by pediatric neuroradiologists in two studies,[13 15] a radiologist plus neurosurgeon in one study[18] and in another pediatric radiologists.[12] There was no description of blinding techniques of those reporting the MRI to the previous imaging undertaken.

Diagnostic accuracy of imaging techniques in included studies

MRI

Pooled analysis for diagnostic accuracy for 52 MRI scans, from two papers,[17 18] compared with surgery found a sensitivity of 97% and a specificity of 94%. There was one false positive where a child had normal findings at surgery and one false negative in an 8-month-old baby later found to have tethering at 2 years 9 months after developing symptoms.

US

Overall heterogeneity of findings was high with an I^2 ranging from 91% to 97%. Meta-analysis of ultrasound diagnosis of clinically relevant SCA diagnoses revealed a sensitivity of 33% and specificity of 87% (see figure 2).

DISCUSSION

The diagnostic accuracy of infant US for clinically relevant SCA for infants with ARM is poor with a sensitivity of 33% and a specificity of 87%. We considered myelomeningocele, tethered cord and lipoma in association with filum terminale thickening clinically relevant and syrinx and isolated lipoma not. The description of

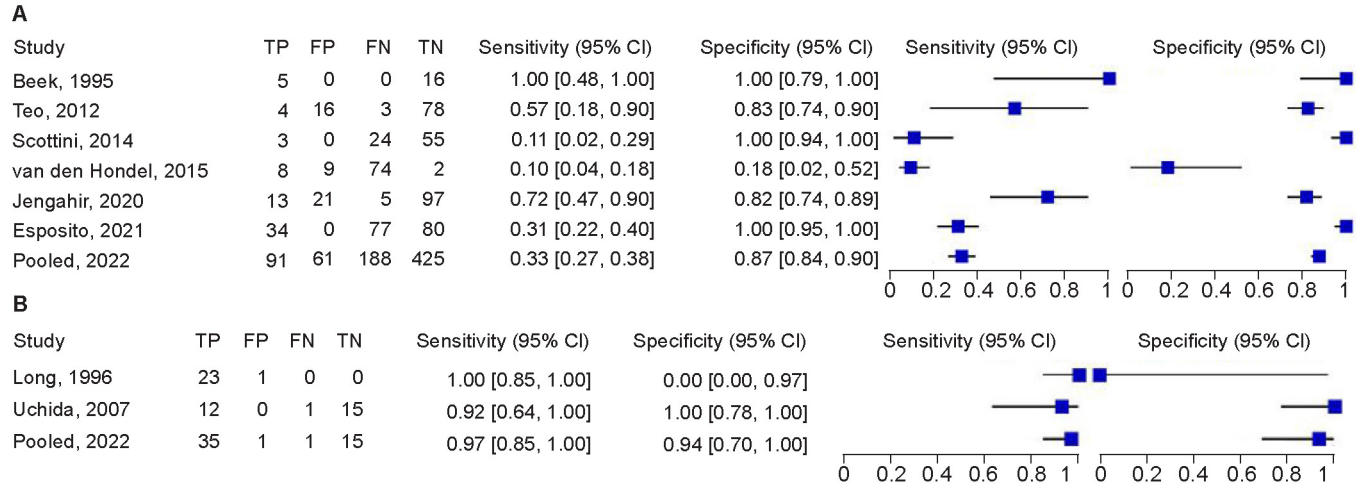

Figure 2 (A) Sensitivity and specificity of ultrasound as compared with MRI for spinal anomalies in children with anorectal malformation (ARM) from included papers and pooled calculations. (B) Sensitivity and Specificity for MRI as compared with surgery for spinal anomalies in children with ARM. FN, false negative; FP, false positive; TN, true negative; TP, true positive.

the findings at US was often at odds with the MRI findings, with US underplaying and missing the significance of the anomaly.[11] Overall, the diagnostic accuracy of MRI for surgically confirmed SCA was much better, with pooled sensitivity of 97% and specificity of 94%.

MRI has been shown to be advantageous over US in other conditions suggestive of an SCA, such as dimples and clefts.[1] If considered as a high risk with a pretest probability of between 20% and 60% cost-effectiveness analysis would also favor MRI.[19] There is an additional increased risk of SCA if the child has another genitourinary anomaly or VACTERL association.[20]

The additional complication to the debate is the role of prophylactic detethering. Bladder improvements are seen in detethering in symptomatic patients,[21 22] studies in children with moderate to severe anorectal malformations have not shown the same impact.[23–25] Children with ARM have bladder dysfunction not exclusively due to the neuropathy caused by a spinal anomaly.[26] The role of neurosurgery in asymptomatic but radiologically defined spinal anomalies is not clear.[27]

Identification of children with an ARM who are at higher risk of neuropathic bladder remains imperative to reduce the risk of renal damage. The authors acknowledge the role of US as a preliminary screening tool to identify those needing combined spinal and urological care early on. But this review highlights how a "normal" infant spine on US does not rule out a clinically relevant SCA.

The methodology of studies in this review, as revealed by Quadas2 analysis, would tend to overestimate the efficacy of US due to the poor protocolized longer term follow-up. We would therefore recommend an MRI spine in all children with an anorectal malformation so that they can be streamed into close clinical follow-up. The decision then is whether the local team feels the risk of a false negative of an early MRI with a feed and wrap approach justifies a general anesthetic in all infants at the age of 12 months. These children already have a continence risk from constipation and surgical intervention, meaning delineating which have a neuropathic bladder is difficult from symptoms alone and are more likely to be missed even with clinical follow-up.

The greatest limitation of our study is the lack of defined follow-up, with general statements being, that as the children did not develop symptoms within the follow-up period, they were considered to have a negative outcome. As the follow-up was poorly described, there may be a tranche of late neuropathic bladders not evident in this paper series. There were few protocolized urodynamic investigations at set time points; therefore, subtle bladder neuropathy may have been missed. Relying on symptom reporting especially in infancy has significant implications of bias. A further limitation is that of those who had an MRI, some were done as screening and others due to symptoms. Only two papers reported the level of anorectal malformation based on the more clinically oriented Krickenbeck classification, which introduces bias when considering the urinary functional prognosis of the patients described. Overall, applicability was reasonable as the papers reflected the population of interest and the outcomes of interest to the clinician. There was no obvious publication bias, but heterogeneity was large.

In conclusion, our results demonstrate the unreliable diagnostic accuracy of spinal US for children with anorectal anomalies. This review supports the recommendation of an MRI spine in all children with ARMs where feasible, to accurately identify those with spinal anomalies and therefore those at higher risk of neuropathic bladder and subsequent renal scarring. Spinal MRI should be a consideration in any multidisciplinary or pediatric urology protocol for safe surveillance of ARM children. The authors veer toward the recommendation of an MRI spine in mid-infancy when feed and wrap is still possible. It is important to recognize that a normal

early infant US spine does not exclude the possibility of an underlying spinal cord anomaly, and this should be considered in light of any developing symptoms.

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