Spinal dysraphism with anorectal malformation: lumbosacral magnetic resonance imaging evaluation of 120 patients

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\textbf{Abstract}

\textbf{Purpose:} We evaluated the prevalence of spinal dysraphism (SD) in patients with anorectal malformation (ARM) by magnetic resonance imaging (MRI).

\textbf{Methods:} From January 2002 to March 2009, 120 patients with ARM who underwent anorectal reconstruction were evaluated for SD with sacral plain film, spinal ultrasonography (US), and lumbosacral MRI. We adopted Krickenbeck international classification of ARM.

\textbf{Results:} Spinal dysraphism was present in 41 (34.2\%) of 120 patients with ARM, 3 (13.0\%) of 23 patients with perineal fistula, 7 (29.2\%) of 24 patients with vestibular fistula, 4 (36.4\%) of 11 patients with rectovesical fistula, 18 (40.9\%) of 44 patients with rectourethral fistula, and 9 (60.0\%) of 15 patients with cloacal anomaly ($P = .04$). Among 41 patients having SD detected by MR, 26 patients (26/41; 63.4\%) underwent detethering surgery for tethered spinal cord. The mean sacral ratio (SR) in patients who underwent detethering surgery for tethered spinal cord was significantly lower than in patients who did not undergo detethering surgery (0.54 ± 0.19 vs. 0.69 ± 0.13; $P < .001$). The optimal cutoff for the SR value predicting SD requiring detethering surgery was 0.605, with sensitivity of 65.4\% and specificity of 77.7\%.

\textbf{Conclusions:} Spinal dysraphism is common in patients with ARM, and its prevalence is higher in patients with complex ARM. Spinal anomalies can occur even with benign types of ARM and, therefore, that all patients should be screened. Magnetic resonance imaging is useful in detecting occult SD that may be missed by conventional radiologic evaluation, physical examination, and spinal US. We further recommend a lumbosacral MRI examination in those whose SR is lower than 0.6.

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The association of anorectal malformations (ARMs) and spinal cord anomalies has been well documented. Recent advances in radiology make it possible for physicians to diagnose spinal dysraphism in patients with ARM more frequently than in the past. Spinal ultrasonography (US) has been considered as an effective screening test. However, magnetic resonance imaging (MRI) is probably the diagnostic modality of choice for evaluating the spine because MRI has excellent resolution of soft tissue of the neuromuscular and skeletal system [1,2]. For example, congenital lesions of the sacrum, including sacral agenesis and meningocele, are optimally imaged with MRI [3]. The results of many studies have described the prevalence of spinal dysraphism in patients with ARM; however, whether the prevalence varies by type of ARM has not been well described, which was noted in only one large retrospective study that has dealt with this topic [4]. Therefore, our objective was to describe the prevalence of spinal dysraphism and tethered spinal cord as diagnosed by lumbosacral MRI in patients with ARM and to determine the prevalence by type of ARM.

1. Material and methods

From January 2002 to March 2009, 142 patients underwent corrective surgery for ARM. As a part of preoperative evaluation, we prospectively performed lumbosacral MRI, regardless of the type of ARM, in all patients who had already been evaluated with plain radiography of the sacrum with or without spinal US. In evaluating the results of plain films of the sacrum, a pediatric radiologist not involved in patient care or follow-up evaluated morphologic abnormalities of the bony sacrum such as dysmorphism, fused vertebra, and hemisacrum. For the purposes of this study, we measured the sacral ratio (SR) in the anteroposterior and lateral sacral films as previously described [5]. Spinal US was performed whenever possible for evaluation of the spinal cord and abnormal mass lesions in the lumbosacral area. The MRI protocol included whole spine sagittal T2-weighted images; lumbar spine axial and sagittal T1-weighted and T2-weighted images; axial and sagittal gadolinium-enhanced T1-weighted images if necessary; and pelvic axial, sagittal, and coronal T2-weighted images when investigation of the pelvic musculature was necessary. In addition, for those who had not been evaluated by MRI previously, we invited the patients to undergo MRI of the lumbosacral area. The pre-MRI sedation protocol consisted of oral chloral hydrate at 100 mg/kg (maximum dose, 2.5 g) given for sedation before MRI, then the patients were transferred to MRI suite with intravenous sedative kit (midazolam and flumazenil). Spinal dysraphism was defined as the abnormalities with imperfect fusion of the midline neural and bony structures. Spinal dysraphism in this study included spina bifida aperta, spina bifida cystica, and spina bifida occulta. Spina bifida aperta was defined as herniation of meninges through the defect of the posterior arch characterized by a non–skin-covered back mass such as meningo(myelo)cele. Spina bifida cystica was defined as herniation of meninges through the defect of the posterior arch characterized by a skin-covered back mass such as lipomeningomyelocele. Spina bifida occulta (occult spinal dysraphism) was defined as other form of spinal dysraphism without back mass (herniation of meninges) such as low-lying conus, tethered spinal cord, spinal lipoma, and anterior sacral meningocele [6]. Neurosurgical operation was recommended for patients with a radiographically demonstrated lipomatous lesion in the filum within the first 6 months of life or, in older children, as soon as the lesion was discovered. Even in those cases, our pediatric neurosurgeons carefully evaluated the presence of symptoms and the size of lipoma before they decide to perform a detethering operation. We did not perform a detethering operation for those who have small, tiny lipomas or asymptomatic low conus medullaris, and who do not want surgery even in those cases with a definite surgical indication. Nonoperative follow-up with biannual examinations was recommended for patients with nonprogressive symptoms or only asymptomatic low conus medullaris. The most notable symptoms in patients with ARM in this study were orthopedic and urologic symptoms. Orthopedic symptoms included progressive scoliosis and cavovarus deformity of the foot with or without leg length discrepancy. Urologic impairment was based on the result of the urodynamic study (neurogenic bladder, vesico-ureteral [VU] reflux). Neurologic symptoms included low extremity muscle weakness and patchy sensory loss. Type of ARM was categorized by the Krickenbeck international classification [7]. We analyzed patient characteristics (age, sex, type of anorectal defect) and prevalence of spinal dysraphism and tethered spinal cord and compared the results of lumbosacral MRI and conventional methods of evaluation such as sacral plain films and spinal US. All patients or their parents (or legal guardians) provided written informed consent for lumbosacral MRI examination of the study. The study was approved by the Yonsei University Graduate School of Medicine Ethics Review Board (Seoul, Korea).

2. Results

During the study period, 120 patients completed lumbosacral MRI evaluation and were enrolled in this study. The remaining 22 patients were not included in this study because we could not obtain consent for MR examination from parents or their guardians during follow-up. There were 72 males (60.0%) and 48 females (40.0%), with a male-tofemale ratio of 1.5:1. Mean ± SD age at anorectal reconstruction was 5.9 ± 10.9 months. The type of ARM included rectourethral fistula in 44 patients (36.7%), vestibular fistula in 24 patients (20.0%), perineal fistula in 23 patients (19.2%), cloacal anomaly in 15 patients (12.5%),
rectovesical fistula in 11 patients (9.2%), and rare or regional variants in 3 patients (2.5%). The mean age at MR evaluation in this study was 3.5 months (range, 1 day to 71.7 months). The results of lumbosacral MRI revealed spinal dysraphism in 41 (34.2%) of 120 patients, including none of the 3 patients (0.0%) with rare or regional variants; 3 (13.0%) of 23 patients with perineal fistula; 7 (29.2%) of 24 patients with vestibular fistula; 4 (36.4%) of 11 patients with rectovesical fistula; 18 (40.9%) of 44 patients with rectourethral fistula; and 9 (60.0%) of 15 patients with cloacal anomaly. Patients with more complex ARM were more likely to have spinal dysraphism ($P = .04$; linear-by-linear association, $P = .001$; Fig. 1A). Among the 120 patients who underwent MRI, 26 patients (21.7%) underwent detethering surgery for the diagnosis of clinically significant tethered spinal cord, including none of the 3 patients (0.0%) with rare or regional variants, in 2 (8.7%) of 23 patients with perineal fistula, 3 (12.5%) of 24 patients with vestibular fistula, 11 (25.0%) of 44 patients with rectourethral fistula, 3 (27.3%) of 11 patients with rectovesical fistula, and 7 (46.7%) of 15 patients with cloacal anomaly. The proportion of patients undergoing detethering surgery was higher in patients with complex types of ARMs, but this did not reach statistical significance ($P = .07$; linear-by-linear association, $P = .001$; Fig. 1B). Low conus medullaris was found in 25 (20.8%) of 120 patients, fatty filum or lipomatous mass lesion in the caudal spine in 24 patients (20.0%), pure dysplastic bony sacrum in 24 patients (20.0%), hydrosyringomyelia in 13 patients (10.8%), dermal sinus tract in 3 patients (2.5%), and meningo(myelo)cele in 2 patients (1.7%) (Fig. 2). Mean ± SD of SR in the 120 patients was 0.65 ± 0.15. The sacral ratio was not significantly different according to the type of anorectal defect ($P = .27$; Fig. 3A). Mean SR measured was 0.76 ± 0.01 in rare or
regional variants, 0.70 ± 0.16 in rectovesical fistula, 0.69 ± 0.13 in patients with perineal fistula, 0.68 ± 0.14 in cloacal anomaly, 0.64 ± 0.16 in vestibular fistula, and 0.62 ± 0.17 in rectourethral fistula. However, mean SR in patients who underwent detethering surgery (0.54 ± 0.19) was significantly lower than in patients who did not undergo detethering surgery. The dotted line for SR of 0.605 represents the cutoff selected from the receiver operating characteristic curve in C. (C) Optimal cutoff SR value from the receiver operating characteristic curve associated with detethering surgery was 0.605 (area under the receiver operating characteristic curve, 0.73 with 95% confidence interval, 0.61-0.85; P < .001) with sensitivity of 65.4% and specificity of 77.7%. CL indicates cloacal anomaly; PF, perineal fistula; RA, rare or regional variant; ROC, receiver operating characteristic; RU, rectourethral fistula; RV, rectovesical fistula; VF, vestibular fistula.

Fig. 3 Sacral ratio in the diagnosis and treatment of ARM. (A) The mean SR did not differ by type of ARM. The dotted line for SR of 0.605 represents the cutoff selected from the receiver operating characteristic curve in C. (B) The mean SR in patients who underwent detethering surgery was significantly lower than in patients who did not undergo detethering surgery. The dotted line for SR of 0.605 represents the cutoff selected from the receiver operating characteristic curve in C. (C) Optimal cutoff SR value from the receiver operating characteristic curve associated with detethering surgery was 0.605 (area under the receiver operating characteristic curve, 0.73 with 95% confidence interval, 0.61-0.85; P < .001) with sensitivity of 65.4% and specificity of 77.7%. CL indicates cloacal anomaly; PF, perineal fistula; RA, rare or regional variant; ROC, receiver operating characteristic; RU, rectourethral fistula; RV, rectovesical fistula; VF, vestibular fistula.

3. Discussion

Evaluation of the sacrum in patients with ARM is important because of the well-established association between sacral hypodevelopment or dysmorphism and poor functional outcome after anorectal reconstruction [5]. In addition to the sacral bony abnormalities, evaluation of spinal dysraphism should include abnormalities of the spinal cord itself because so-called spinal cord tethering is associated with various orthopedic [8-10], urologic [8,11-13], and gastrointestinal problems [14,15]. In the present study, the

Fig. 4 depicts spinal US and MRI findings in a patient with occult spinal dysraphism.
prevalence of spinal dysraphism in patients with ARMs was 34.2%, and clinically significant spinal cord tethering requiring detethering surgery was 21.7%, consistent with previous studies [16-25]. More complex types of anorectal defects were associated with a higher prevalence of spinal dysraphism. Our data have more statistical power than previous series [18,19,26], with the largest number of patients with ARM studied, all of whom underwent MRI. It is likely, therefore, that the results of our study estimate the true prevalence of spinal dysraphism in patients with ARM and confirm that prevalence of tethered spinal cord increases as the severity of the anorectal defect increases. The results of several studies showed even higher prevalence of tethered spinal cord among patients with low type of ARM when MRI was routinely used to evaluate the spinal cord [2,25,27], compared with the results of the present study (8.7% of patients with perineal fistula). One possible explanation for this difference may be different practice patterns for the diagnosis of spinal cord tethering requiring detethering surgery. Tethering of the spinal cord should be assessed with care to determine whether it is true cord tethering or an incidental finding of low conus medullaris. No relationship was observed between the type of ARM and the coexistence of anomalies in bony sacrum expressed either qualitatively (sacral morphology) or quantitatively (SR). Sacral abnormalities frequently did not predict the presence of abnormal intraspinal lesions such as fatty filum, spinal cord tethering, and clinically important syrinx (Fig. 2). Measurements of sacral morphology in the newborn period may be inaccurate because of incomplete ossification. The results of lumbosacral MRI usually shows the cause of the tethering and provides sufficient anatomical details of the lesion. Occult
spinal dysraphism was observed in 22.1% of patients who
did not show spinal dysraphism on sacral plain film and
spinal US, which is consistent with the results of the study of
Long et al [28] who found potentially important spinal
dysraphic anomalies in 21% of patients with an apparently
normal spine. These results emphasize the need to rule out
dysraphic anomalies even in patients with no detectable
vertebral anomalies. Taken together, MRI is the ideal method
for evaluating the spinal cord in patients with ARMs. An
additional benefit of MRI is the ability to diagnose remnant
urticular structures and genitourinary malformations such as
megacystis, ureteral anomalies, and horseshoe kidney. It also
has the ability to illustrate anatomical details of the levator
muscles, all of which are very important in the perioperative
management of patients with ARM. Measurement of the SR
was suggested by Peña [5] for establishing functional
prognosis with reasonable accuracy in patients with ARM.
Its value in normal children was 0.74. Warne et al [29] found
high variability of SR values among similar patients,
suggesting that a single result has limited value in
discriminating a normal from an abnormal sacrum. Further-
more, in another study, SR was not predictive of inconti-
ence after anorectal reconstruction [30]. Nevertheless, the
results of the present study suggest that SR is useful for
predicting the presence of tethered spinal cord in patients
with ARM, which accords with the results of the study by
Peña [4]. The sacral ratio in patients who underwent
detethering surgery was significantly lower than in patients
who did not undergo detethering surgery, and we were able
to determine an optimal cutoff for the SR from the receiver
operating characteristic curve with moderately high levels of
sensitivity and specificity. Thus, we believe that SR is
valuable in detecting clinically important spinal cord
tethering that requires detethering surgery. Considering
cost-effectiveness, we recommend MRI for patients with
both sacral hypodevelopment and SR below 0.6. The results
of spinal US may be inaccurate. As a major referral center
for the treatment of ARM, many children are referred to us at
several months of age after the sonographic window of the
spine has closed. This may be especially true for toddlers
with previously undiagnosed perineal fistula (so-called
anterior anus). As a matter of fact, for considerable number
of patients with ARM including the patients who had been
referred to us after the age when the sacral window for spinal
US was closed, the accuracy of the spinal US was
disappointing, and often, the examination itself was
impossible. Therefore, we changed our policy not to perform
spinal US for all patient with ARM from the latter half of the
period. A total of 33 of 120 patients underwent spinal US at
the period of first half of study (median age, 51 postnatal
days; ranged from 0 to 230 postnatal days). The data in
Table 1 were based on our own observations about the
correlation between spinal US and MRI in 33 patients. In our
experience, screening US was acceptable because of high
specificity and positive predictive value. However, sensitiv-
ity and negative predictive value were rather low. The
problem in our study was that US had a relative higher false-
negative rate when compared to the previous reports.
Ultrasonography is safe, noninvasive, inexpensive, does
not require sedation, and can be performed portably [31].
Furthermore, the correlation between spinal US and MRI has
been excellent, and spinal US is currently accepted as an
initial screening tool for detecting spinal dysraphism in
patients with ARM [1,32-34]. Therefore, it is not to be
denied that US is an ideal screening tool for evaluating the
spine in patients with ARM. However, our data showed more
discordance rate between US and MRI than previous results.
We suppose that false-negative rate of US was high because
(i) US could not accurately detect small lipoma or fatty filum
in the spinal canal. In this study, MRI was used as a standard
tool to detect spinal dysraphism, and it is possible that this
method has ability to diagnose minimal types of spinal dysraphism that might
not be evident on US. The significance of minimal types of
spinal dysraphism detected by MRI remains to be proved in
the future because it is obviously abnormal finding and has
potentials to grow over time. (ii) Transitional vertebrae were
the potential for confusion over the labeling or assignment of
vertebral levels during evaluation of the tethered spinal cord
in US. (iii) The experience of sonographer and the time
interval between US and MRI might affect the outcomes.
Borg et al [35] performed screening spinal US in 24 patients
with “high” type ARM found that spinal US finding was
equivocal in about 21% (5/24) of the patients. Furthermore,
false-negative result of the spinal US was found in 1 of the 14
patients who underwent MR imaging of the spine. Davidoff
et al [19] performed screening MRI in 44 patients (including
21 former patients who were asymptomatic) and remarked
that at least 9% (4/44) of the patients had significant occult
spinal dysraphism that underwent neurosurgery. All of this
amounts to saying that the difference of data seems to be a
matter of method or technique for examination. The view is
widely held that MRI is the gold standard for evaluation of
spine and spinal cord [1,2,36,37]. However, US is still
accepted as an excellent screening tool in the initial

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<th>An analysis of US findings according to age of the patients, below and above 3 months</th>
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<td>MRI</td>
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<td>Age ≤ 90 days</td>
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<td>US</td>
<td>Normal 13</td>
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<td>Spinal dysraphism 0</td>
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<td>Total 13</td>
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<td>Age &gt; 90 days</td>
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evaluation because US has many advantages as described above. The results of US in our study are somewhat different from those of previous studies, though it remains to be proved in the future. The benefit of detethering surgery is also controversial. The results of many studies suggest that neurosurgical release of tethered spinal cord has a beneficial effect on urinary [9,12,38,39], orthopedic [9], and bowel function [39]. Evidence of such a beneficial effect was not clearly observed in bowel function in the present study. For example, we observed that the patients with rectovesical fistula having spinal dysraphisms detected by MRI universally showed poor outcomes whether detethering surgery was done. In those patients, a bowel management program including antegrade continence enema procedure was necessary. One possible explanation is because an animal experiment showing that neurologic pathways controlling urinary and fecal continence may be established during early embryogenesis [40]. Another important aspect of spinal cord tethering is that it is not a static condition, with several reports of progressive neurologic deficit in children with tethered cord syndrome [38,41]. The present study includes one patient who showed newly developed tethered cord syndrome based on the results of serial MRI, which is similar to the previous report [9]. These patients have an anatomically stable but growing lipomatous mass because of increased lipogenesis within the spinal canal during the early years of life. Our study has several limitations. The present study may be said to have skewed data because of the racial or ethnic diversity of ARM or spinal dysraphism that show different levels of occurrence. The study population was from a referral center, which tends to overestimate the prevalence of conditions compared to a true population-based study sample. As far as the functional benefit of the detethering surgery is concerned, we do not have satisfactory data yet and the follow-up after detethering surgery is still short. Although the decision for neurosurgical intervention for tethered spinal cord was based on symptoms and findings in the lumbosacral MRI, for most patients, urologic and orthopedic symptoms were not aggravated, and bowel function seemed unchanged after detethering surgery. Considering that symptoms of tethered spinal cord can be reliably evaluated after children reach school age or often become more significant after a growth spurt in previously unaffected patients, more long-term follow-up is necessary. Furthermore, as the criteria for neurosurgical intervention can be varied according to the practice patterns, the operative findings in this study cannot be used a gold standard for deciding whether patients have clinically important spinal dysraphism. In conclusion, spinal dysraphism has a high prevalence among patients with ARM, especially with complex ARM. Spinal anomalies can occur even with benign types of ARM, and therefore, all patients should be screened. Results of MRI of the lumbosacrum in our study are very useful in detecting spinal dysraphism in these patients, especially occult spinal dysraphism that may be missed by conventional radiologic evaluation, physical examination, and spinal US. With these points in mind, we think it is reasonable to recommend patients with ARM having sacral hypodevelopment with SR value below 0.6 to undergo a MRI of the lumbosacral area to evaluate for tethered spinal cord. Although it is well known that MRI is more accurate than US in the evaluation of spinal dysraphism and US is more useful than MRI in the initial screening in patients with ARM, high false-negative rate of the spinal US in our study casts doubt on the role of spinal US to screening tool with MRI being reserved for cases where spinal US is equivocal or has revealed a definite abnormality. However, as the accuracy of our US data is currently limited because of a variety of reasons as mentioned above, it remains to be proved in the future.

References


