Low-Risk Lumbar Skin Stigmata in Infants: The Role of Ultrasound Screening

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Objective To reassess the utility and validity of ultrasound (US) screening in infants with lumbar midline skin stigmata (MSS) that may be associated with tethering of the spinal cord.

Study design We conducted a prospective observational study of 254 infants under age 6 months with suspicious dorsal MSS between 2005 and 2007. All infants were examined by US and neurosurgical clinical evaluation, and 50 infants also underwent magnetic resonance imaging (MRI). The US and MRI findings were analyzed for correlation. Associations between the imaging findings and the presence of the low-risk skin lesions simple dimple (113 cases) and deviated gluteal fold (DGF; 44 cases) also were evaluated.

Results Analysis of US and MRI results for the cohort of 50 neonates in whom both examinations were performed showed high concordance. The low-risk group of infants with simple dimple and DGF constituted 157 US procedures, 96% of which were of high quality, providing clear visualization of spinal components. None demonstrated any clinically significant pathological findings.

Conclusions Our data reaffirm the reliability of US as a screening tool for tethered cord syndrome. Infants with low-risk lesions, such as simple dimple and DGF, may be absolved from US screening, because these findings alone do not indicate underlying pathological lesions. We propose a simplified diagnostic classification system for MSS. (J Pediatr 2009; - - - -).

It is well known that tethered cord syndrome (TCS) is associated with dorsal midline skin stigmata (MSS).1-3 Early detection of TCS is important because prophylactic cord release, when indicated, prevents most if not all symptoms;4-8 however, the majority of infants with MSS have normal underlying anatomy. Thus, differentiating between skin lesions according to the degree of association with cord tethering is crucial to guide appropriate management. There is a lack of consensus regarding the classification and diagnostic workup of infants with MSS. Although ultrasound (US) provides a reliable, noninvasive, easily accessible screening tool, it is operator-dependent and limited by vertebral ossification relative to age. Magnetic resonance imaging (MRI) is the gold standard for diagnostic screening, but it is costly and requires general anesthesia in this age group.9-11 Many attempts have been made to clarify this diagnostic challenge. The absence of standardized MSS nomenclature further hinders a systematic discussion of this issue.11-13

Although there is a low incidence of TCS in neonates with simple dimple and deviated gluteal fold (DGF), the optimal diagnostic workup for these infants remains unclear.14,15 In the present study, we focused on these low-risk lesions, examining the role of, validity of, and need for high-quality US examination in affected infants. We propose a simple classification system and corresponding imaging guidelines for neonatal MSS.

Methods

This was a prospective observational study that included data from clinical and imaging examinations obtained according to standard clinic procedures, in adherence to Good Clinical Practice Guidelines and as approved by the Sourasky Medical Center’s Institutional Review Board. Consecutive infants up to age 6 months referred either directly for US screening or for neurosurgical consultation due to suspicion of MSS by a primary care physician were included. Each examining physician completed the relevant parts of a standard questionnaire. Data were compiled using the File Maker Pro database for statistical analysis.

Infants were assessed clinically, including a complete neuro-orthopedic examination. MSS was classified according to the following predetermined criteria by the first physician to examine the patient. A midline dimple was defined as “simple” if
it presented as a soft tissue depression appearing up to 2.5 cm above the anus or within coccygeal proximity, regardless of subjective impressions of its size, depth, and trajectory. The term “pilonidal sinus” was avoided, because it is a misleading, nondescriptive etiologic term that may be mistakenly reassuring. All other dimples were considered “nonsimple.” DGF included all cases of abnormal folds, whether bifid, split symmetrical, or any other degree of deviation, without an underlying mass. Discolorations included vascular anomalies, hemangiomas, pigmented lesions, and cutis aplasia. Mongolian spots were categorized separately and considered low-risk skin stigmata.1,3,4,6 Hypertrichosis defined cases with long, silky hair, as opposed to low-risk light hair. Additional categories included pendeculated skin tag (often referred to as human tail), fibroma pendulum, and any midline protrusions, including lipomas, categorized as a midline mass (Figure 1). These were considered high-risk stigmata.1,3,4,6

All patients were referred for US examination. A 2-dimensional linear probe, at 7 to 10 MHz, was used to scan the spine with the patient in the prone position with the spine flexed over a pillow, to accentuate the lumbosacral joint. Images were obtained of the lumbosacral junction (to identify vertebra L5), sagittal images up to the mid-thoracic spine, and axial images, thereby allowing analysis of cauda equine symmetry and cord pulsatility (Figure 2).

The following US findings, highly indicative of cord tethering, were considered abnormal: low height of the conus medullaris (below the L2-L3 intervertebral disc) and normal height of the conus medullaris (L2 vertebrae and above), accompanied by any of the following: syrinx, intrathecal mass or lipoma, dermal sinus (ie, a dorsal tract transversing the soft and bony tissue, ending above the cul de sac of the subarachnoid space, continuous with intraspinal structures), thickened terminal filum (> 2 mm or with a bulbous appearance), or absence of cord pulsatility. The following findings were considered inconclusive: borderline height of the conus medullaris at the L2-L3 intervertebral disc, isolated finding of hyperechoic terminal filum (of normal thickness), and dorsal or ventral displacement of the cord.

US quality was graded on to a 3-level scale as high (all spinal elements could be clearly viewed), good (all required assessment criteria could be analyzed but with a lower degree of clarity), or poor (only the essential assessment criteria [ie, skeletal structure, height of the conus medullaris, and presence of masses] could be analyzed).2,5,16,17

In a subset of 50 infants, MRI was performed using a 1.5 T MRI scanner (Sigma Horizon, Echo speed, LX MRI scanner; GE Healthcare, Milwaukee, Wisconsin). For this examination, the neonate was placed in the supine position under general anesthesia. T1- and T2-weighted sagittal, axial, and coronal images at least up to the thoracic spine, with image slice thickness ranging from 2.5 to 5 mm, were obtained.

**Statistical Analysis**

The McNemar test and kappa index were used to establish levels of concordance between MRI and US images of the various skin lesions. The $\chi^2$ test or Fisher exact test, according to sample size, were applied to analyze correlations between various dorsal skin lesions and the radiologic findings.

**Results**

Prospective, observational data were gathered on 254 infants (127 females and 127 males) under age 6 months with suspicious dorsal MSS. Simple dimple was the most common lesion in these infants (n = 125), followed by DGF (n = 53). US examinations were completed at a mean age of 7 weeks (standard deviation ± 5 weeks). The subset of neonates with isolated DGF was examined on average at a later age of 9 weeks (± 6 weeks).

Of the 254 infants in the study group, 50 underwent MRI examination after US, at an average age of 20 weeks. Although many of the infants referred for MRI were lost to follow-up, this cohort of 50 infants allowed a significant

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**Figure 1.** A, Neonate with a simple dimple < 25 mm above the anus. B, Neonate with a symmetrical DGF. C, Neonate with an underlying mass resulting in deviation of the gluteal fold. Such cases were classified according to the underlying lesion.
comparison of concordance between MRI and US. In the analysis of the value of the height of the conus medullaris, borderline L2-L3 intervertebral disc height was considered abnormal. In the analysis of normal versus abnormal overall evaluation of US and MRI, 3 inconclusive US studies (2 cases due to borderline height and 1 case due to an echogenic filum of normal thickness) were excluded because of inability to define the findings as normal or abnormal. These 3 cases were included in the statistical analysis comparing each of the imaging criteria, as single variables. Table I compares the US and MRI findings, demonstrating excellent sensitivity (96%), specificity (96%), and positive predictive value (96%) regarding the overall final evaluation.

Any minor discrepancies between US and MRI were not statistically significant. There was 1 case of a false-negative US, in which the US was assessed as normal and the MRI revealed a small terminal syrinx. There also was 1 case of a false-positive US, in which the US indicated a dermal sinus and the MRI showed the tract to be of insignificant height, below the thecal sac, and thus normal. The US examinations also revealed 2 cases of borderline conus height that were identified as normal by MRI.

Although US had high specificity for all criteria, it had low sensitivity for dermal sinus (46%). Six cases of dermal sinus were identified on MRI but not on US. This lack of US sensitivity did not affect appropriate management, however; all of the infants with dermal sinus missed by US had skin lesions that were more severe than those of the low-risk group, and thus of less relevance to our low-risk MSS analysis.

### Table I. Comparison of US and MRI findings

<table>
<thead>
<tr>
<th>Radiologic criterion</th>
<th>US Normal MRI</th>
<th>Abnormal MRI</th>
<th>McNemar P value*</th>
<th>US sensitivity</th>
<th>US specificity</th>
<th>US positive predictive value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Skeletal assessment</td>
<td>Normal</td>
<td>39</td>
<td>1</td>
<td>1</td>
<td>91%</td>
<td>100%</td>
</tr>
<tr>
<td>Conus height†</td>
<td>Normal</td>
<td>33</td>
<td>0</td>
<td>0.233</td>
<td>100%</td>
<td>94%</td>
</tr>
<tr>
<td>Filum thickness</td>
<td>Normal</td>
<td>31</td>
<td>1</td>
<td>0.198</td>
<td>92%</td>
<td>97%</td>
</tr>
<tr>
<td>Thick fatty filum</td>
<td>Normal</td>
<td>45</td>
<td>0</td>
<td>1.00</td>
<td>100%</td>
<td>98%</td>
</tr>
<tr>
<td>Lipomyelomeningocele</td>
<td>Normal</td>
<td>41</td>
<td>0</td>
<td>1.00</td>
<td>100%</td>
<td>98%</td>
</tr>
<tr>
<td>SCM</td>
<td>Normal</td>
<td>48</td>
<td>0</td>
<td>1.00</td>
<td>100%</td>
<td>100%</td>
</tr>
<tr>
<td>Dermal sinus</td>
<td>Normal</td>
<td>36</td>
<td>6</td>
<td>0.344</td>
<td>46%</td>
<td>99.6%</td>
</tr>
<tr>
<td>Syrinx</td>
<td>Normal</td>
<td>44</td>
<td>2</td>
<td>0.5</td>
<td>66%</td>
<td>100%</td>
</tr>
<tr>
<td>Ventriculus terminalis</td>
<td>Normal</td>
<td>40</td>
<td>6</td>
<td>0.07</td>
<td>67%</td>
<td>85%</td>
</tr>
<tr>
<td>Final evaluationz</td>
<td>Normal</td>
<td>23</td>
<td>1</td>
<td>0.644</td>
<td>96%</td>
<td>96%</td>
</tr>
</tbody>
</table>

SCM, split cord malformation.

*Significance of discrepancy between US and MRI.
†Borderline conus height was considered abnormal for the purpose of comparison.
zNote that the statistical analysis of the overall final evaluation does not include 3 cases of inconclusive US.
Simple Dimple

Overall, there were 125 cases of simple dimple; of these, 93 cases were isolated, 20 cases were combined with other low-risk lesions (light down, Mongolian spots, and/or DGF), and the remaining 12 cases were combined with more severe lesions. In this study we focused on cases of isolated simple dimple and simple dimple in combination with low-risk lesions. Of these 113 cases, 4 cases had US images of poor quality and thus were excluded from the analysis, minimizing operator dependency, leaving a total of 109 cases. In this group, 8 MRIs were performed (Table II).

US images of the 90 isolated cases of simple dimple analyzed exhibited no distinctive pathologic findings. There was 1 false-positive US, with the US image indicating an echogenic filum revealed as normal on MRI. A single inconclusive US indicating a borderline (L2-L3 intervertebral disc) height of the conus was not followed by MRI because of parental preference.

Among the 19 cases of simple dimples combined with other low-risk skin lesions, 18 US studies were normal. A single inconclusive US indicating a normal conus height with echogenic filum was not followed by MRI because of parental preference.

MRI was performed in 8 infants. In 7 of these 8 cases, the reason for the MRI was parental or surgeon preference. All 8 MRIs were normal.

Overall, all of the US studies of good quality in this group (n = 109) were normal, except for 1 case of borderline conus height. No infants had a dermal sinus; 20 infants had insignificantly low dermal tracts, below the cul de sac of the subarachnoid space with no continuity with intraspinal structures.

Deviated Gluteal Fold

Overall there were 53 cases of DGF, including 32 isolated cases and 12 cases with other low-risk lesions (ie, light down, Mongolian spots, and/or simple dimple). Of the remaining 9 cases, 8 cases were combined with more severe lesions and 1 case was a patient with non random association of birth defects: vertebral anomalies, anal atresia, cardiovascular, tracheoesophageal fistula, renal/radial anomalies, limb anomalies (VACTER) syndrome. Of 44 cases of isolated DGF and those combined with low-risk lesions, the US images for two isolated DGF cases were of poor quality and thus were excluded from analysis, minimizing operator dependency, leaving a total of 42 cases. In this group, 2 MRIs were performed (Table II). In all cases in which DGF appeared as an isolated finding, US revealed no pathological findings. A single inconclusive US indicating a borderline height of the conus was not followed by MRI because of parental preference. Among the 12 cases of DGF in combination with other low-risk skin lesions, all 12 US studies and the 2 MRIs performed were normal. Overall, all of the 42 US examinations of good quality in this group were normal, except for a single inconclusive case of borderline conus height. No infants had a dermal sinus; 5 infants had insignificantly low dermal tracts (below the cul de sac of the subarachnoid space) with no continuity with intraspinal structures.

Discussion

Comparing images in the group of 50 infants with both US and MRI examinations allowed us to establish the reliability of US as a screening tool for TCS. We found that US had excellent sensitivity (96%), specificity (96%), and positive predictive value (96%) overall compared with MRI. Individual imaging variables also demonstrated high concordance. Even when the US seemed to indicate a false-negative or a false-positive result, this led to no alterations in management. The slight discrepancies in conus height between the MRI and US images may be due to the narrow anatomy of the L2-L3 intervertebral disc. The absence of false-negative US assessments reaffirms the reliability of US despite the technical difficulties involved. US may have a low sensitivity for dermal sinus detection yet still achieve a high specificity. All cases of dermal sinus missed by US were linked to more severe skin lesions than those seen in the low-risk group. Thus, we conclude that for cases of low-risk MSS, US of optimal quality can serve as a highly reliable screening tool to rule out cord tethering abnormalities. This conclusion confirms preliminary opinions that have been previously published by others.

Our definition of simple dimple includes previously used terms (pilonidal sinus and sacrococcygeal dimples) and precludes the need to determine dimple size, depth, and trajectory, which is difficult to do in the clinical practice and is of minimal significance to further management. Within our

Table II. Imaging findings of simple dimple and DGF

<table>
<thead>
<tr>
<th>Skin lesion</th>
<th>Imaging mode</th>
<th>Skeletal assessment</th>
<th>Conus height</th>
<th>Filum thickness</th>
<th>Overall assessment</th>
<th>Surgery</th>
</tr>
</thead>
<tbody>
<tr>
<td>Simple dimple</td>
<td>US (n = 109)</td>
<td>109 normal</td>
<td>108 normal, 1 borderline</td>
<td>109 normal</td>
<td>106 normal, 3 inconclusive</td>
<td>0</td>
</tr>
<tr>
<td>MRI (n = 8)</td>
<td>8 normal</td>
<td>8 normal</td>
<td>8 normal</td>
<td>8 normal</td>
<td>8 normal</td>
<td></td>
</tr>
<tr>
<td>DGF</td>
<td>US (n = 42)</td>
<td>42 normal</td>
<td>41 normal, 1 borderline</td>
<td>42 normal</td>
<td>41 normal, 1 inconclusive</td>
<td>0</td>
</tr>
<tr>
<td>MRI (n = 2)</td>
<td>2 normal</td>
<td>2 normal</td>
<td>2 normal</td>
<td>2 normal</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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group of 109 infants with simple dimple, US revealed no pathological findings indicating the need for surgery. In this group, we found 20 cases of low dermal tracts, below the cul de sac of the subarachnoid space, with no continuity with intraspinal structures. As such, these dermal tracts were unrelated to cord tethering and necessitated no additional evaluation or surgery.\(^\text{15}\)

We conclude that cases of simple dimple, either isolated or in combination with other benign (ie, low-risk) lesions, do not necessitate MRI. In fact, according to our data, simple dimple is not associated with tethering of the spinal cord. Therefore, infants with simple dimple do not require US imaging. If in a given infant a question arises regarding the stigmata classification, then US can serve as a reliable screening tool for TCS.

In our group of 42 infants with DGF or split gluteal fold, US revealed no distinctive pathological findings. Five infants exhibited low dermal tracts, considered insignificant. Thus, we conclude that infants with DGF and split gluteal fold, either isolated or in combination with other benign (ie, low-risk) lesions, do not require MRI. When the deviation is caused by an underlying soft or bony mass, or when DGF is part of a greater systemic syndrome (eg, VACTER), MRI is indicated.

Previous studies have assessed similar variables and the association between skin lesions and cord tethering. Schropp et al\(^\text{18}\) studied the level of association and correlation between skin lesions and pathological findings in 358 patients between age 1 day and 8 years who required surgery and found an association between DGF and lipoma, placing DGF in a high-risk group. But in these cases, the anatomical deviation itself was most probably caused by the underlying lipoma. In our study, such cases were categorized as masses separate from DGF, thereby resolving the apparent discrepancy regarding risk stratification, as described later.

Kriss and Desai\(^\text{19}\) prospectively analyzed 207 newborns with suspicious skin lesions, including 160 with isolated simple dimple, all of whom had a normal US evaluation. Our data reaffirm the findings of this group, testing the validity of their findings at a later age and adding further information regarding DGF management.

Guggisberg et al\(^\text{14}\) attempted to develop management guidelines based on an evaluation of 54 patients age 2 days to 16 years with various types of skin lesions. In these patients, there were only 3 cases of isolated simple dimple and 5 cases of DGF, all of whom had a normal US. These investigators attempted to absolve simple dimple from US screening, based on previous reports, but recommended US screening for DGF.\(^\text{19}\) Our study clarifies the difference between cases of innocent DGF with no association with TCS and cases of DGF with an underlying mass, occurring in association with a systemic syndrome, or both, which are more likely to require surgery.

In our prospective study, all US examinations were performed by a single senior pediatric radiologist (L.B.S.). The quality of and conclusions that can be drawn from US under such conditions may be strongly operator-dependent. Inexperienced examiners may increase the rate of false-positive and false-negative findings. US of the lumbar spine must be performed at a young age and by a professional imaging expert trained in the field. In addition, open communication between the radiologist and clinician is crucial to effective evaluation.

The present study focuses on simple dimple and DGF in the low-risk group. At the other end of the spectrum are the well-recognized high-risk skin stigmata that should routinely prompt MRI evaluation. Between these polarities lies the gray zone of intermediate-risk skin stigmata that necessitate further research to define the roles of US and MRI. We suggest the following risk group stratification and imaging recommendations for suspicious dorsal MSS in infants under age 6 months. (Cases with multiple skin stigmata are stratified according to the lesion of highest risk.) In the low-risk group (ie, simple dimple, split gluteal fold or DGF, Mongolian spots, or light hair), the need for imaging is questionable. If imaging is desired, US provides reliable screening when performed by an experienced pediatric radiologist. (Note that when the deviation is caused by an underlying soft or bony mass, or when DGF is part of a greater systemic syndrome, such as VACTER, the stigmata are classified as high risk.) In the intermediate-risk group (any midline lumbar discoloration), whether a normal US is sufficient to obviate the need for MRI remains unclear. In the high-risk group (ie, nonsimple dimple, hypertrichosis, tags, or any mass), MRI is always indicated.

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