Skin Markers of Occult Spinal Dysraphism in Children

A Review of 54 Cases

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Objectives: To verify the diagnostic value of lumbosacral midline cutaneous lesions in asymptomatic children to detect occult spinal dysraphism (OSD) and to propose a practical approach for clinical investigations with respect to the type of cutaneous lesions observed.

Design: Retrospective study of 54 children referred to the Department of Pediatric Dermatology between 1990 and 1999 for congenital midline lumbosacral cutaneous lesions.

Setting: The private or institutional practices of participating dermatologists and pediatricians.

Main Outcome Measures: Evaluation of the diagnostic value of midline cutaneous lesions for the detection of OSD. Association of skin examination findings with spinal anomalies detected by magnetic resonance imaging or ultrasound.

Results: Occult spinal dysraphism was detected in 3 of 36 patients with an isolated congenital midline lesion and 11 of 18 patients with a combination of 2 or more different skin lesions.

Conclusions: A combination of 2 or more congenital midline skin lesions is the strongest marker of OSD. Careful dermatologic examination is needed to detect suggestive markers and request a spinal magnetic resonance image, which is the most sensitive radiologic approach to detect an OSD.

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Spinal dysraphism refers to a spectrum of congenital anomalies characterized by an incomplete fusion of the midline mesenchymal, bony, or neural elements of the spine. As skin and nervous tissue are of ectodermal origin, anomalies of both may occur simultaneously. Occult spinal dysraphism (OSD) is characterized by skin-covered lesions without exposed neural tissue.

Congenital midline paraspinal lesions, mostly localized in the lumbosacral area, are widely recognized as markers of OSD. They include subcutaneous lipomas, dermal sinuses, tails, and localized hypertrichosis. In the presence of such cutaneous lesions, radiologic investigations must be performed to detect a possible OSD. Hyperpigmented lesions and aplasia cutis congenita are also reported to be associated with OSD. The screening value of isolated lumbosacral vascular lesions remains difficult to determine because, despite the International Society for the Study of Vascular Anomalies (ISSVA) classification, ambiguity persists in the terminology used in the literature. At present, the importance of dimples is still under discussion. Furthermore, no one has established whether isolated deviation of the gluteal furrow (DGF) represents a cutaneous marker of OSD.

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In the present retrospective study, we report the occurrence of OSD in a series of 54 children referred for lumbosacral midline cutaneous lesions. In all patients, spinal radiologic investigations were performed to detect an OSD. Taking into consideration the data from this series and a review of the literature, we propose a practical approach for clinical diagnosis of OSD focusing on the type of cutaneous lesions observed.

We reviewed the clinical records of pediatric patients who presented with congenital mid-
line lumbosacral skin lesions and/or isolated DGF who were referred to the Department of Dermatology at Hôpital Necker-Enfants Malades, Paris, France, between 1990 and 1999 (dermatologic consultation register). Patients with obvious vertebral spinal lesions such as myelomeningocele were not considered (as they had been observed in the Department of Neurosurgery since birth).

The medical records of all affected patients, including neonatal data and inpatient and outpatient records, were reviewed by 2 experienced dermatologists (D.G. and D.H.-T.) who used standardized forms to collect clinical information and investigation results. When necessary, obstetric and pediatric medical records were also consulted, as were specialists from other disciplines who followed up with the patients.

Magnetic resonance images (MRIs) and ultrasound images (USDs) were examined by both a radiologist (F.B.) and a neurosurgeon (M.Z.). Examples are shown in Figure 1 D-G.

Hamartoma is a term, coined from the Greek word meaning “to err,” used to refer to a tumorlike but primary nonneoplastic malformation showing an abnormal mixture of normal tissue components. The lesion may be abnormal in its topography or the amount of different tissues.

Hemangioma (Figure 2A) and port-wine stain (PWS; Figure 2B) were diagnosed according the ISSVA classification. Hypertrichosis corresponds to an excess of normal hair with a lumbosacral topography. An acrochordon is a soft pedunculated “skin tag” and can be categorized as either a tail or a fibroma pendulum. Tail was further categorized into either a human tail (neurectodermal appendage, Figure 2C) or a faun tail (congenital midline lumbosacral tuft or patch of abnormal hair, Figure 2D).

RESULTS

Between 1990 and 1999, 54 children (37 girls, 17 boys) were examined. The mean age at consultation was 2.3 years (range, 2 days–16 years). All the patients had a dermatologic examination performed by an experienced dermatologist (Table 1). Neurologic and/or orthopedic anomalies were clinically detected in 3 patients (patients 3, 4, and 5; Table 2).

The observed congenital midline lumbosacral cutaneous lesions and the rate of their association with an OSD are reported in Table 1. Thirty-six patients (67%) presented with an isolated lumbosacral cutaneous lesion, and 18 patients (33%) presented with a combination of 2 or more lesions: 11 patients had 2 lesions; 6 patients had 3; and 1 patient had 4 lesions. The most frequently occurring cutaneous lesions, isolated or in combination, were PWS (n = 26) and DGF (n = 15). Lipomas were
also frequently found (n=11). Dermal sinuses were noted in 5 patients; they presented as a punctum in all 5 patients and were never explored during consultation (ie, they were not drained nor was depth determined). For all 5 of them, the characterization was obtained after neurosurgery and is reported in Table 2 for 4 patients. The fifth patient had an obstructed fistula limited to the dermis.

All patients underwent spinal radiologic examination. Fifty-one patients underwent spinal MRI, 22 of these before age 6 months. In 14 cases, MRI detected an OSD (Table 2). Spinal USD was performed in 15 patients and always before age 6 months. In 13 cases, the lumbosacral USD was performed prior to the MRI (patients 4-14; Table 2; data not shown for remaining 2 patients). Of these 13 patients, 7 had normal USDs, although spinal MRI findings were abnormal (patients 5-9, 13, and 14; Table 2). Both explorations showed abnormal findings in 4 cases (patients 4, 10, 11, and 12; Table 2). Both explorations showed normal findings in 2 patients. In 2 cases, only spinal USD was performed, and it was considered normal. One patient had normal findings on a spinal computed tomographic scan. These 3 last patients each had an isolated midline lesion (PWS, fibroma pendulum, and lipoma, respectively).

For the 13 patients who underwent both spinal MRI and USD, MRI was performed for the following reasons: (1) Many of these patients had 2 or more congenital lumbosacral cutaneous lesions (patients 4-11 and patient 14; Table 2; data not shown for 1 patient with normal results from both explorations). (2) Two patients had very suggestive isolated lesions (patients 12 and 13; Table 2). (3) One patient had associated DGF and cleft lip and palate; nonetheless the MRI and USD findings were normal.

Occult spinal dysraphism was detected in 14 patients (26%; Table 2). An isolated lesion was associated with OSD in 3 patients (patients 4-11; Table 2). Patients 12 and 13 presented with a dermal sinus and a lumbosacral lipoma, respectively. Patient 14 presented with 2 unclassified hamartomas, each associated with an underlying OSD (Figure 1C and G). The other isolated lesions (PWS, hemangioma, DGF, hypertrichosis, simple dimple, fibroma pendulum, and pigmentary nevus) were not associated with OSD. Among the 18 patients with a combination of 2 or more congenital midline cutaneous

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Figure 2. Clinical aspects of isolated or combined congenital median lumbosacral cutaneous lesions. A, Ulcerated hemangioma centered on a dermal sinus and deviation of the gluteal furrow; B, isolated port-wine stain; C, human tail; D, faun tail.
lesions, 11 (20%) had an OSD (Table 2). Lipoma with overlying PWS (n=5) and DGF (n=6) were the most frequent lesions associated with OSD. Spinal lipoma (n=10) and tethered spinal cord (n=9) were the most frequent spinal anomalies (Table 2).

Many cutaneous lesions associated with OSD have been reported.3,28-30 It is important for these skin markers to be recognized to detect underlying spinal anomalies. Published data have shown that about 76% of patients with OSD (range, 43%-95%) have single or combinations of median or paramedian congenital lumbosacral cutaneous lesions.3-7,15,16 Herein we report our experience in a retrospective study of 54 cases.

A combination of 2 or more congenital midline skin lesions constitutes the strongest marker of OSD. In our series, 11 of the 18 patients with such a combination had OSD. In consequence, very careful dermatologic examination is required, giving importance to all features such as discrete DGF.

Isolated lipomas or lipomas occurring in combination with other lesions, mostly with an overlying PWS, are the most common midline cutaneous lesions associated with OSD.3,31 Our data confirm the previous publications (found in 7 of 14 patients). Deviation in the gluteal furrow is a reliable associated feature (3 cases; Table 2 and Figure 1A-B and D-F).

Two patients presented with tails in combination with other lesions. Both had OSD (Table 2). Faun tails appear as abnormal tufts or patches of hair (Figure 2D).22 Human tails are more rarely described and seem to correspond to the persistence of a caudal vestigium, which normally develops and disappears at the end of the first month of the embryonic phase (Figure 2C).23-27 These skin alterations are not well defined by physicians and are mostly treated surgically, for purely aesthetic reasons, without any search for an underlying OSD. By consequence, tails are often not detected before neurologic clinical manifestations.11,22-25,32,33

In our study, a dermal sinus was associated with OSD in 4 of 4 patients (Table 1; Figure 1B and Figure 2A). All 5 patients underwent surgical exploration because dermal sinuses carry a high risk of cerebrospinal fluid infection or intradural abscesses.31,34 One patient had only an obstructed fistula limited to the dermis. In a series of 28 patients with dermal sinus, 27 had uncharacterized skin

| Table 1. Congenital Midline Lumbosacral Cutaneous Lesions and Their Association With Occult Spinal Dysraphism Encountered in 54 Patients
<table>
<thead>
<tr>
<th>Type of Cutaneous Lesion</th>
<th>Isolated Lesion (n = 36)</th>
<th>Associated Lesions* (n = 18)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Port-wine stain</td>
<td>16</td>
<td>10</td>
</tr>
<tr>
<td>Hemangioma</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>DGF</td>
<td>5</td>
<td>10</td>
</tr>
<tr>
<td>Hypertrichosis</td>
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<td>3</td>
</tr>
<tr>
<td>Dimple</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>Fibroma pendulum</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>Pigmentary nevus</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Lipoma</td>
<td>4</td>
<td>1</td>
</tr>
<tr>
<td>Unclassified hamartoma</td>
<td>1†</td>
<td>1</td>
</tr>
<tr>
<td>Dermal sinus</td>
<td>1‡</td>
<td>4</td>
</tr>
<tr>
<td>Tail‡</td>
<td>0</td>
<td>2</td>
</tr>
</tbody>
</table>

Abbreviations: DGF, deviation of the gluteal furrow; OSD, occult spinal dysraphism.

*Associated lesions means more than 1 midline skin lesion (eg, DGF and lipoma) in the same patient.
†The same patient (patient 14; Figure 1C and G) presented with 2 hamartomas (1 dorsal and 1 lumbar) that were associated with 2 different spinal lipomas.
‡Concerns both human and faun tails (Figure 2C and D).
changes. In this report, Ackerman and Menezes emphasized the importance of characterization of congenital midline skin markers in detection of OSD.

None of the 5 patients who presented with a “simple” dimple had OSD. We defined simple dimple, or coccygeal pit, as an isolated small dimple (≤5 mm in diameter) 2.5 cm or closer to the anus and localized just above the gluteal furrow. In a recent series of 207 neonates with 216 cutaneous stigmata, Kriss and Desai found that midline simple dimple was the most common skin lesion (74%). The authors differentiated between the simple dimple (which is not a cutaneous marker of spinal dysraphism) and atypical dimple (which is highly associated with underlying spinal dysraphism): 8 (40%) of 20 patients with atypical dimples were found to have OSD. Atypical dimples seemed to be large (≥5 mm), high on the back (≥2.5 cm from the anus), and appeared in combination with other lesions. Radiologic screening is warranted in cases of atypical dimples. In a series of 75 neonates with sacral simple dimple, Gibson et al reported normal spinal USD findings. Hence, radiologic screening is not advisable in cases of isolated simple dimple.

In the present study, isolated hypertrichosis was not associated with OSD. In only 1 case among 3 of hypertrichosis in combination with other lesions was OSD found. Hypertrichosis is reported as a marker of diastematomyelia. In a series of 43 cases of diastematomyelia, Miller et al noted that 17 patients had hypertrichosis above the spinal dysraphism. In 6 cases, hypertrichosis was associated with other cutaneous lesions. Kein and Greene reported hypertrichosis in 40% of patients in a series of 112 cases of diastematomyelia. In our opinion, the thick abnormal hair overlying diastematomyelia is more like a faun tail than like hypertrichosis (Figure 2D).

In our series, we distinguished between PWS and hemangioma according to the ISSVA classification. All patients with PWS and hemangioma were evaluated by MRI. None of the 16 patients with isolated PWS showed an OSD (Table 1; Figure 2B). In 7 of 10 cases of PWS in combination with other lesions, detection of OSD was positive. Occult spinal dysraphism was absent in 2 cases of PWS with DGF and 1 case of PWS with simple dimple. The clinical aspect of PWS did not differ between the 2 groups. Neither of the 2 patients with isolated hemangioma had OSD. On the other hand, OSD was present in 2 of the 3 patients who had hemangioma in combination with other lesions.

Because ISSVA classification is not used in all publications, conclusions about the detection value of congenital midline vascular lesions for OSD remain difficult to draw, and more studies are required. Some authors have concluded that medial telangiectatic vascular nevi in the sacral region, also called “butterfly-shaped marks,” are benign lesions and do not entail further diagnostic investigations in the absence of signs or symptoms. No imaging data are available in this article. More recently, Allen et al performed MRI or USD in 20 patients with lumbosacral strawberry nevi. The 4 patients with underlying OSD also had other cutaneous stigmata. However, the description of strawberry nevi was insufficient to classify according to ISSVA.

In a report of 5 patients with lumbosacral hemangioma combined with other developmental anomalies, Goldberg et al considered such hemangiomas as cutaneous markers of OSD (Figure 2A). Albright et al described 7 neurologically normal infants with lumbosacral hemangiomas, all of whom had a tethered spinal cord. However, 5 of them also had other skin tags.

Regarding published data, isolated PWS does not seem to be associated with OSD. As a marker of OSD, PWS is more frequently described in a combination of midline lumbosacral cutaneous lesions. Classically, a DGF is not considered a cutaneous marker of OSD. However, an apparently isolated DGF may be the only visible sign of an underlying subcutaneous lipoma. In the present study, 15 patients had a DGF (Figure 1A and B and Figure 2A). It was isolated in 5 cases, and MRI did not detect an underlying OSD. In 10 cases, the DGF was combined with other lesions, mostly lipoma and/or PWS, and in 6 of these, MRI revealed OSD. Only a prospective study including a sufficient number of patients could determine whether a strictly isolated DGF warrants screening for OSD.

In our series, we did not find midline aplasia cutis congenita to be a sufficient finding to seek consultation. Some observations of aplasia cutis congenita and lumbosacral hyperpigmented lesions associated with OSD have been reported. The exact diagnostic significance of midline lumbosacral aplasia cutis congenita is controversial. In cases of pigmented lesions or unclassified hamartoma (Figure 1C), a precise semilologic and/or histopathologic analysis fails in most cases. Consequently, the exact nature of the lesion cannot be ascertained (nevus, lentigo, or “cafe au lait” spots?), and analysis of the results is therefore quite difficult.

Magnetic resonance imaging is a noninvasive and nonirradiant examination. It is now considered to be the first-choice examination for the detection of OSD. By MRI, the intraspinal extension of a lipoma and the exact localization of the conus medullaris, both of which affect surgical possibilities, can be ascertained (Figures 1D-G). Also, MRI can preempt the use of myelography, which is not completely risk free. Nevertheless, MRI requires sedation in the newborn and in small children.

High-resolution spinal USD allows quick and noninvasive evaluation of an OSD in the newborn. It does not require any premedication, and it costs less than MRI. Ultrasound can be applied easily and constitutes a good means of screening newborns suspected of OSD because the posterior elements of the spine are nonossified in the neonate before age 5 to 6 months. The entire spinal cord must be explored because the skin defects do not always precisely overly the spinal dysraphism. Eleven of our 14 patients with OSD underwent both USD and MRI (Table 2, footnote). In 7 of the 11 cases, the USD findings were normal, while MRI detected an OSD. Similar discordance has been reported elsewhere. Interpretation of USD is indeed difficult and operator dependent. Spinal computed tomography was performed in only 1 patient, and conclusions about its screening value in detecting OSD are quite limited. Thus, in the presence of cutaneous lesions highly suggestive of dysraphism, and particularly in the presence...
of a combination of cutaneous lesions, spinal MRI must be performed to rule out OSD even when previous USD findings were normal. Radiographic evaluation is nearly useless in detection of OSD.

Finally, patients may be categorized into 3 groups (Table 3). The first group comprises patients with a high risk of OSD. It includes patients with 1 or more of the following: (1) a combination of 2 or more congenital midline lumbosacral skin lesions of any kind; (2) only 1 congenital midline cutaneous lesion in association with spinal cord dysfunction such as genitourinary and/or rectal problems (eg, recurrent infections or incontinence), abnormal gait, or abnormal arching or ulcers of the feet; or (3) only 1 isolated but very suggestive congenital midline cutaneous lesion such as a lipoma, with or without an overlying PWS, dermal sinus, or tail. Magnetic resonance imaging must be performed to detect OSD even when previous USD findings were normal. In particular, dermal sinuses are associated with a high risk of cerebrospinal fluid infection leading to rapid spinal MRI and neurosurgical evaluation.\(^{31,34}\)

The second group comprises patients with isolated lesions that carry a lesser risk of OSD: aplasia cutis congenita, unclassified hamartoma, DGF, and atypical dimple. Nonetheless, evaluation for OSD should be performed for patients in this group. In children younger than 6 months, it may be acceptable to propose first a spinal-screening USD performed by an experienced radiologist. Magnetic resonance imaging may be performed after doubtful or abnormal USD findings. In children 6 months or older, MRI is necessary to exclude OSD.

The last group comprises patients with isolated lesions that are not usually associated with OSD: PWS, hemangioma, hypertrichosis, simple dimple, pigmentary nevus, and mongolian spot. In such cases, a radiologic investigation does not seem to be appropriate. Isolated PWS can be compared with occipital nevus flammeus, present in about 40% of newborns. Nevus flammeus has no pathologic significance.

Detection of OSD necessitates regular neurologic and neurosurgical follow-up to investigate any spinal cord dysfunction, mostly in locomotion and sphincter function. Any manifestation detected should prompt multidisciplinary consultation to arrive at the appropriate treatment for each patient. Nevertheless, surgical treatment of medial lumbosacral cutaneous lesions for purely aesthetic reasons, without prior careful exclusion of an associated spinal lesion, must be avoided.

In conclusion, OSD is frequently associated with a combination of 2 or more different congenital midline lumbosacral lesions. It is important to detect these lesions before the occurrence of neurologic or orthopedic manifestations. Magnetic resonance imaging is the best radiologic imaging method. However, USD may be used in some cases. A few isolated lesions are clearly associated with OSD: lipomas, dermal sinuses, and tails. Isolated PWS does not seem to be of pathologic significance, and further investigation is not required. Appreciation of the diagnostic value of aplasia cutis congenita, isolated pigmentary nevus, and isolated DGF is more difficult. The presence of such lesions seems to require a systematic search for OSD.

**Table 3. Assessment of Congenital, Medial Lumbosacral Cutaneous Lesions in the Absence of Neurologic or Orthopedic Manifestations⁰**

<table>
<thead>
<tr>
<th>Risk of OSD</th>
<th>Congenital Lumbosacral Midline Skin Lesion</th>
<th>Age &lt; 6 mo</th>
<th>Age ≥ 6 mo</th>
</tr>
</thead>
<tbody>
<tr>
<td>High</td>
<td>&gt;2 Lesions of any kind</td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>One lesion and spinal cord dysfunction</td>
<td></td>
<td>MRI</td>
</tr>
<tr>
<td></td>
<td>Lipoma††</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Tail††</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Dermal sinus††</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Group 1</td>
<td>MRI</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>One lesion and spinal cord dysfunction</td>
<td></td>
<td>MRI</td>
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<tr>
<td></td>
<td>Lipoma††</td>
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<td></td>
<td>Tail††</td>
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<tr>
<td></td>
<td>Dermal sinus††</td>
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</tr>
<tr>
<td>Group 2</td>
<td>USD</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Normal Abnormal</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Deviation of gluteal furrow††</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>No investigations</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>MRI</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Group 3</td>
<td>No investigations§</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>PWS</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Hypertrichosis</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Pigmentary nevus</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Simple dimple†</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Mongolian spot</td>
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</tbody>
</table>

Abbreviations: MRI, magnetic resonance imaging; OSD, occult spinal dysraphism; PWS, port-wine stain; USD, ultrasound.

*Neurologic and morphologic evaluations are required in the presence of clinical symptoms.
†Considered as isolated lesion.
‡High risk of cerebrospinal fluid infection leading to rapid spinal MRI and neurosurgical evaluation.
§All authors do not agree with this opinion for isolated PWS.
\(\)Simple dimple is defined as an isolated small lesion (≤5 mm in diameter) 2.5 cm or closer to the anus.

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