Sacral Dimples

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Author Disclosure
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Objectives

After completing this article, readers should be able to:

1. Explain the difference between open and closed neural tube defects.
2. Describe the characteristics of spinal skin dimples that warrant further evaluation.
3. Describe the characteristics of spinal skin dimples that do not warrant further evaluation.
4. Discuss the evaluation of spinal skin dimples and name the findings that suggest occult spinal dysraphism.
5. Discuss the neurosurgical treatment of occult spinal dysraphism.
6. Explain the natural history and clinical manifestations of occult spinal dysraphism.

Definitions

Neural tube defects are among the most common forms of birth defect, affecting 1 in every 1,000 pregnancies. (1)(2) These defects, which result from abnormal fusion of the neural tube during embryonic development, are placed into two broad categories: open and closed. Open neural tube defects are lesions in which brain, spinal cord, or spinal nerves are exposed through obvious defects of the meninges and skull or vertebral column. Examples are anencephaly, myelomeningocele, and meningocele. Closed neural tube defects are skin-covered lesions under which the nervous system structures have not formed normally. These include split cord malformation, dermal sinus tract, tethered spinal cord, and intraspinal lipoma (Table).

Spina bifida is an imprecise term often used to describe a variety of congenital spinal anomalies that range in consequence from insignificant to severe. Spina bifida occulta (SBO) is a radiographic finding that describes incomplete osseous fusion of the posterior elements. It may occur in conjunction with a cutaneous abnormality but is clinically benign and is considered a normal variant. (3) Occult spinal dysraphisms (OSDs) are much less common than SBO and encompass a variety of skin-covered neural tube defects. Because the neural structures are affected, however, neurologic impairment is common. Most forms of OSD have an associated overlying cutaneous abnormality.

Most open neural tube defects are diagnosed prenatally with ultrasonography and serum marker concentrations. Those defects not identified before delivery are apparent at birth. An OSD, on the other hand, is less obvious and may not be diagnosed until later in life, despite its presence at birth. The occult nature can be problematic because the clinical impairments associated with closed neural tube defects, which include paresis, spasticity, sensory disturbance, orthopedic deformity or contracture, and bowel and bladder dysfunction, often progress insidiously over time.

Diagnosis

More than 50% of OSDs are diagnosed when a dimple (Fig. 1) is noted in the lower spine/sacral region. Although the natural history of OSD is not fully understood, early diagnosis and intervention are believed to improve outcome in almost all cases. (4) Hence, the recognition of a suspicious skin dimple and identification of underlying anomalies with prompt radiographic evaluation and neurosurgical referral is crucial. However, not all dimples are associated with an OSD. Distinguishing between cutaneous stigmata associ-

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Table. **Definitions**

Open Neural Tube Defects	
Anencephaly	An exposed rudimentary brainstem due to failed closure of the cephalic portion of the neural tube.
Myelomeningocele	Herniation of the spinal cord through an unfused portion of the spinal column.
Meningocele	Protrusion of the meninges through a spinal column opening.
Closed Neural Tube Defects	
Split Cord Malformation Diastematomyelia	Division of the spinal cord into two parts that are usually separated by bone or cartilage.
Dermal Sinus Tract±Inclusion Tumor	Incomplete dysjunction (separation of the cutaneous ectoderm and neuroectoderm), resulting in an epithelial-lined tract that terminates in neural structures.
Tethered Spinal Cord	Abnormal attachment of the lower end of the spinal cord to surrounding structures.
Lipomyelomeningocele or Spinal Cord Lipoma	Premature dysjunction allows mesodermal infiltration between cutaneous ectoderm and neuroectoderm, resulting in a tethered spinal cord attached to a benign fatty tumor in the back.

ated with OSD and innocent skin dimples can be difficult and may lead to costly and unnecessary tests or referrals. (5) Therefore, the focus of this review is to provide information on how to identify skin dimples that require further evaluation, what method of evaluation should be used, and when to refer to a specialist.

Most cutaneous stigmata associated with OSD are found in the midline overlying the spinal lesion. A finding of hypertrichosis, capillary hemangioma, atretic meningocele, subcutaneous mass (eg, lipoma), or a caudal appendage (Fig. 2) requires further investigation. Gluteal cleft anomalies other than dimples also have a weak association with milder forms of OSD and warrant further evaluation. Therefore, a deviated or duplicated ("split") gluteal cleft (Fig. 3) should raise concern for OSD, whether or not a dimple is present. The management of a "dimple" alone, however, demands greater diagnostic acumen because some dimples over the spine represent dermal sinus tracts, although most do not. Dermal sinus tracts are not classified as open neural tube defects because characteristically they do not feature overt meningeal, osseous, and cutaneous defects (despite potential communication between the skin and nervous elements). These skin-covered lesions are marked similarly to other OSDs with overlying dimples or other cutaneous stigmata. (6)

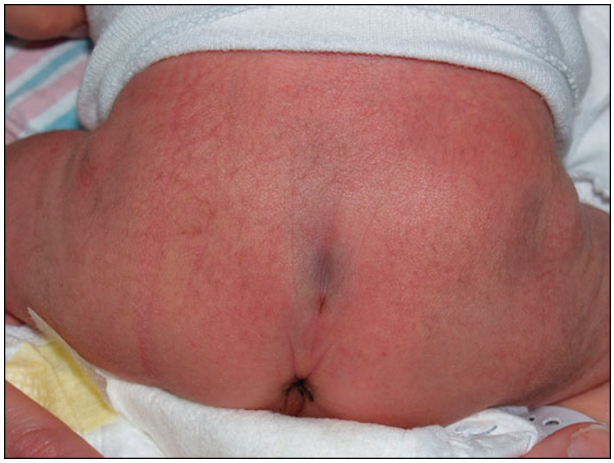

Figure 1. Solitary dimple whose location greater than 2.5 mm above the anus indicated the need for further evaluation, which revealed an occult spinal dysraphism requiring neurosurgical intervention.

Clinical findings do not predict with certainty which dimples are associated with OSD. However, the following criteria have been found to differentiate best between dimples that require further evaluation and those that require only routine follow-up evaluation: multiple dimples, dimple diameter larger than 5 mm, location greater than 2.5 cm above the anal verge, and association of the dimple with other cutaneous markers. Review of the literature shows that 2% to 4% of all children have a dimple identified in the sacrococcygeal region, but only seven cases have ever been found to be associated with an OSD. (7) For those patients in whom a coccygeal dimple was found in conjunction with an OSD, most had more than one dimple. Notably, the second dimple often was found more rostrally along the spine (ie, cervical, thoracic, or lumbar). Hence, clinical examination should seek to identify dimple location and the number present.

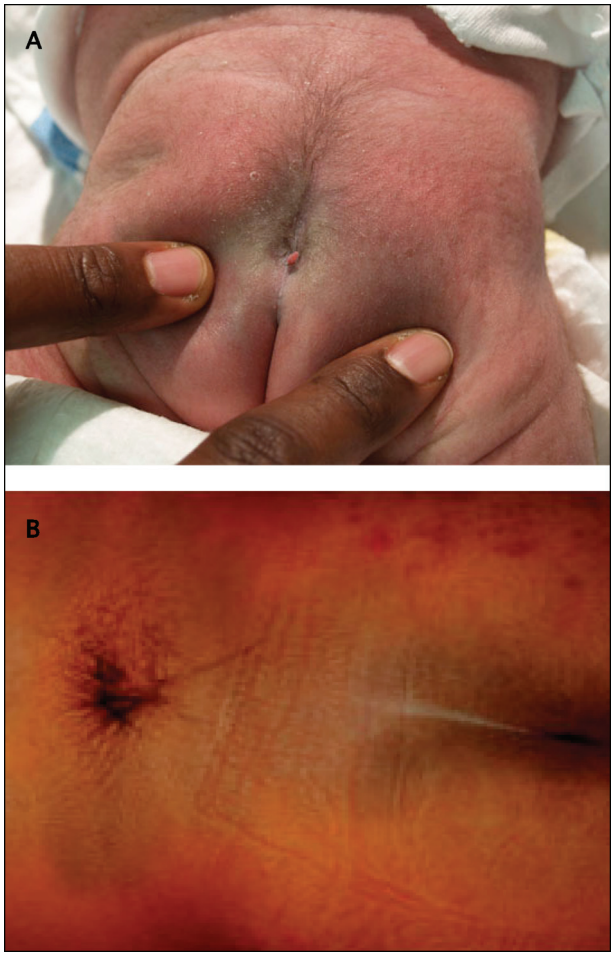

Figure 2. Cutaneous lesions. Skin dimples are often the cutaneous marking found with occult spinal dysraphism. However, multiple other markings can signify underlying spinal element malformation, including caudal appendage (A) and hypertrichosis (B).

Solitary sacrococcygeal pits located entirely within the gluteal cleft (Fig. 4) have no clinical significance and should be considered anatomic variations of normal. Typically, the coccyx is palpable beneath the dimple and intact skin can be seen at the base (Fig. 5). If there is difficulty discerning whether the lesion is covered completely by skin, otoscopic examination of the dimple often can determine if there is a bottom to the pit. Although most lesions occur in the midline, eccentric lesions (Fig. 6) are not in themselves abnormal unless occurring in conjunction with other lesions or outside the sacral spine. No radiographic evaluation or neurosurgical consultation is required; parental reassurance is the only intervention necessary.

In addition to a thorough inspection of the skin, the pediatrician must perform a careful physical examination, with particular attention to the neurologic and orthopedic aspects. Associated orthopedic findings can include clubfeet, arthrogryposis (contracture of multiple joints leading to fixation of the joints in extension or flexion) of the lower extremities, and hip dislocation. Abnormal curvature of the spine, including kyphosis or scoliosis, also may be present. Abnormal neurologic or orthopedic examination findings indicate the need for further evaluation.

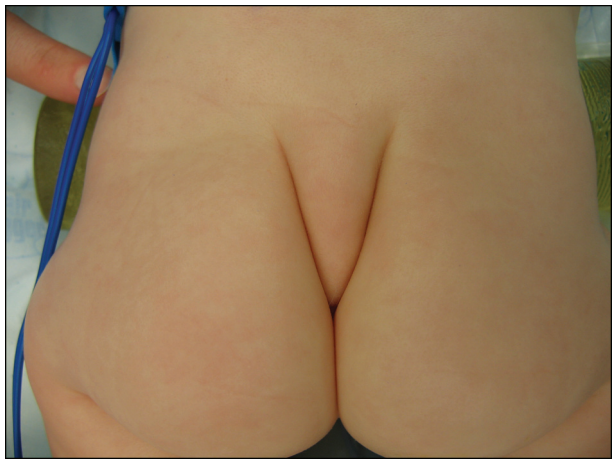

Figure 3. A duplicated gluteal cleft associated with occult spinal dysraphism.

Management

When detailed history and physical examination raise the clinical suspicion for OSD, radiographic imaging should be obtained (Fig. 7). Either ultrasonography or magnetic resonance imaging (MRI) can be employed to evaluate

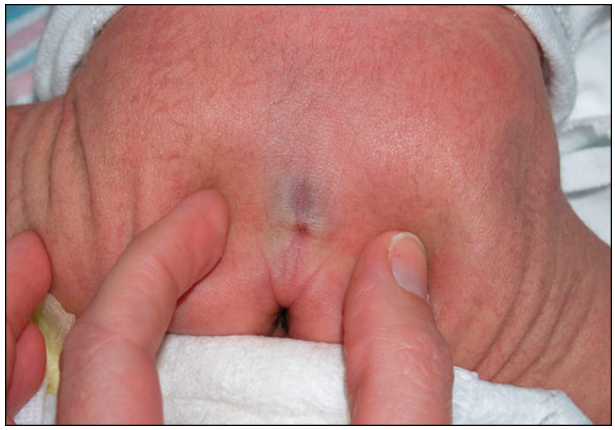

Figure 4. A prototypical benign sacral dimple that is located within the gluteal cleft (less than 2.5 cm above the anus) and solitary.

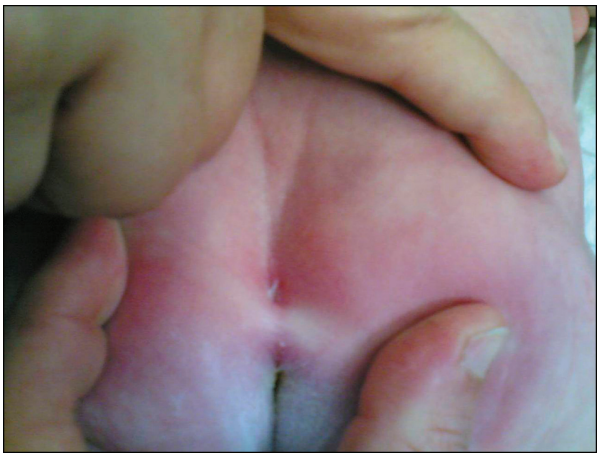

Figure 5. Solitary sacrococcygeal dimple that demonstrates complete covering with skin over the entire dimpled area when the skin is stretched laterally and, therefore, is not an occult spinal dysraphism–associated lesion.

OSD. Ultrasonography of the lumbosacral spine generally is useful only in children younger than 3 months of age because ossification of the vertebral arches has not yet occurred. (8) However, the decision to use ultrasonography versus MRI (for children of any age) as first-line imaging appears somewhat institution-dependent. In one study of a pediatric population who had sacrococcygeal cutaneous lesions, a discordance rate of 17% between ultrasonography and MRI studies was found in which ultrasonography suggested an OSD while MRI yielded normal results. (9) Pediatricians, therefore, should be aware of the possible discrepancy in findings with these imaging modalities and know which study is most appropriate at their respective institutions.

Spinal ultrasonography can assess the level of the conus medullaris, the diameter and echogenicity of the filum terminale, and the position and movement pattern of the spinal cord and nerve roots. Abnormal findings can include a low-lying conus, in which the tip is below the level of the second lumbar vertebral body; a filum terminale diameter greater than 2 mm; and a posteriorly positioned or nonmobile cord, which can indicate tethering. If ultrasonographic findings are abnormal, MRI of the spine is indicated. Findings on MRI vary, based on the type of OSD present. In general, MRI is more reliable and exact in diagnosing OSD.

Neurosurgical referral is appropriate if radiographic evaluation reveals any spinal abnormality. Consideration for early referral (before imaging) is appropriate for dimples superior to the gluteal cleft, especially if any discharge is observed or reported. Such dimples are the

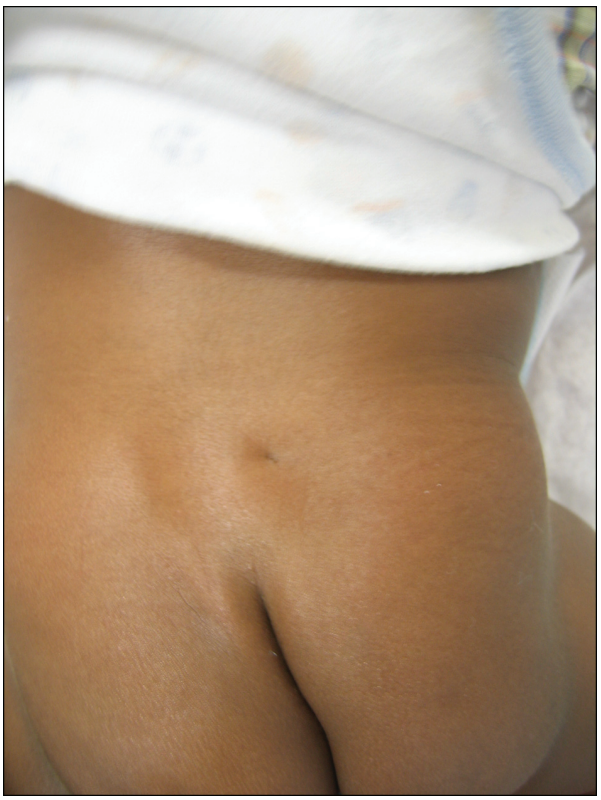

Figure 6. A right eccentric dimple that occurs outside of the midline but does not carry a high degree of suspicion for an occult spinal dysraphism because it is an isolated sacrococcygeal lesion.

hallmark of dermal sinus tracts that predispose the patient to bacterial meningitis or intraspinal abscess. (10) Surgical intervention is aimed at untethering the spinal cord and removing abnormal tissue, when present.

Prognosis

Almost all neurosurgical referrals for suspected OSD in children younger than 1 year of age are for evaluation of a dimple. Although the natural history of OSD is somewhat unpredictable, the overall risk of neurologic compromise increases with time. Neurologic deficits can be difficult to identify in young children because the onset of dysfunction is generally insidious and occurs about the same time as expected neurologic function development (eg, crawling, walking, standing). Accordingly, OSD deficits may be mistaken for delayed accruement of normal function, and irreversible damage may occur before symptomatic manifestation. The reasons for neurosurgical referral for children older than 1 year of age suspected of having OSD include chronic urinary tract infections, lower limb deformity (eg, foot drop,

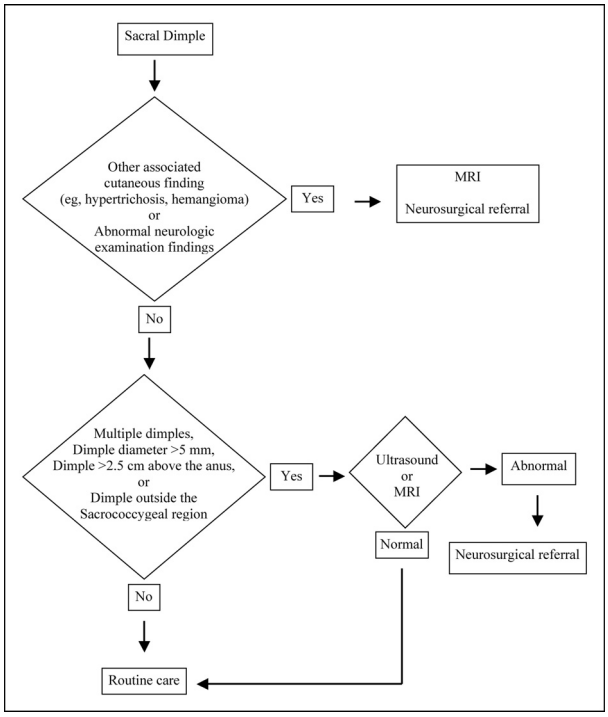

Figure 7. An algorithm for evaluation of dimples overlying the neural axis. MRI=magnetic resonance imaging

weakness or atrophy in a lower extremity, talipes equinovarus, or dragging one foot), bowel/bladder dysfunction, pain, and lower extremity spasticity or paresis. However, careful inspection of this population often reveals subtle cutaneous stigmata. Therefore, it is important for the pediatrician to be vigilant in searching for midline skin anomalies. Even as common a condition as primary nocturnal enuresis warrants careful examination for midline skin anomalies.

Conclusion

Early diagnosis of OSD often comes from identification of spinal skin dimples. Recognition of suspicious lesions is important to reduce the risk of neurologic, urologic, and orthopedic dysfunction. During examination, the pediatrician should not only look for dimples along the spine but also for other markings such as abnormal hair growth, asymmetric gluteal creases, dermal sinuses/dimples/pits, hyper- or hypopigmentation, capillary hemangiomas, skin tags, and subcutaneous fatty masses that are associated with OSDs. Any lesion along the spine outside of the sacrococcygeal region or identification of more than one skin marking anywhere along the spine warrants further evaluation, including radiographic imaging and neurosurgical referral. (11) Optimal outcome is most likely with early diagnosis and surgical intervention.

Summary

- Spinal skin dimples and other cutaneous markings located outside of the sacrococcygeal region are associated most often with closed neural tube defects or OSD.
- The presence of more than one skin dimple anywhere along the neural axis is an indicator of the likely presence of OSD.
- The neurologic deficits associated with OSD are progressive and frequently not detected until permanent dysfunction has been sustained when diagnosed later in life.
- Early neurosurgical intervention is believed to prevent or halt progression of neurologic deficits due to spinal cord tethering.

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PIR Quiz

Quiz also available online at http://pedsinreview.aappublications.org.

11. Which of the following is the best example of an open neural tube defect?

    A. Anencephaly.
    B. Dermal sinus tract.
    C. Diastematomyelia.
    D. Spinal cord lipoma.
    E. Tethered spinal cord.

12. Among the following, the child *most* likely to benefit from early referral to a neurosurgeon is:

    A. 1-month-old who has an eccentric sacral dimple.
    B. 1-week-old who has a solitary sacrococcygeal pit.
    C. 2-month-old who has a sacrococcygeal dimple.
    D. 3-month-old who has a dimple superior to the gluteal cleft with discharge.
    E. 3-week-old who has a palpable coccyx beneath the dimple.

13. An 8-month-old girl presents to your clinic with multiple dimples superior to the gluteal cleft, and you suspect OSD. Among the following, the *most* appropriate next step in her evaluation is:

    A. Computed tomography scan of the spine.
    B. Lumbar puncture.
    C. Magnetic resonance imaging of the spine.
    D. Ultrasonography of the spine.
    E. Radiographs of the spinal column.

14. A father brings his 6-year-old son to you for evaluation of nocturnal enuresis and occasional daytime wetting. On physical examination, you note a sacral dimple. Among the following, the feature *most* concerning for OSD is:

    A. Eccentric sacral location of the dimple.
    B. History of one urinary tract infection.
    C. Hypertrophy of one foot.
    D. Spasticity of the lower extremities.
    E. Truncal hypotonia.

15. Of the following, the feature that *best* distinguishes a dimple associated with OSD is:

    A. Cutaneous marker associated with the dimple.
    B. Greater than 3 mm maximal dimension.
    C. Location greater than 1 cm above the anus.
    D. Sacrococcygeal location.
    E. Single dimple.