Occult Spinal Dysraphism

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Spinal dysraphism is an incomplete fusion of the spine during embryologic development and encompasses a spectrum of congenital anomalies that can affect the spinal cord, nerve roots, and vertebral column. These anomalies can be categorized into open and closed spinal dysraphism. Open spinal dysraphism is also known as spina bifida aperta. The neural tissue in these lesions is exposed, and examples include meningoceles and myelomeningoceles. Closed spinal dysraphism is also referred to as occult spinal dysraphism (OSD). In these lesions, the neural tissue is covered with skin, making the diagnosis less apparent. Isolated vertebral defects are the most common and least severe form of OSD. Other examples of OSD include neurenteric cysts, split notochord syndrome, split spinal cord malformation, sacral meningeal cysts, spinal lipomas, caudal regression syndrome, dorsal dermal sinus tracts and cysts, and tethered cord syndrome. Among these diagnoses for OSD, evaluations for potential tethered cord syndrome and dermal sinus tracts are the most common reasons for referrals to pediatric neurosurgeons. Because the scope of this *In Brief* cannot go into detail with all of these diagnoses, we describe tethered cord syndrome and dorsal dermal sinus tracts in more detail and then focus on the epidemiology, associated morbidity, physical findings, challenges with diagnosis, and management of all types of OSD.

Tethered cord syndrome can result from several types of spinal dysraphism. A tethered cord arises when the spinal dysraphic malformation creates a situation where the spinal cord is anchored in some way, thereby causing the spinal cord to stretch as the child grows. This leads to ischemic changes in the neurons. Findings on imaging depend on the type of spinal dysraphism involved, but generally, concern about a tethered cord is heightened if the conus medullaris terminates caudal to the L2 to L3 disc space or if the diameter of the filum exceeds 2 to 3 mm. Children with tethered cord syndrome can develop progressive motor and sensory signs and symptoms, including gait abnormalities, muscle atrophy, abnormal deep tendon reflexes, loss of bladder control, lumbosacral pain, orthopedic deformities of the foot, and scoliosis.

Dorsal dermal sinus tracts are communications between the surface of the skin and deeper tissues. The sinus tracts are lined by stratified squamous epithelium and are associated with lumbosacral dimples or pits. They may terminate in the subcutaneous tissues or extend into the subarachnoid space and attach to the lower spinal cord, resulting in an intradural extension. Intradural extensions place patients at risk for meningitis because bacteria can enter the subarachnoid space via the sinus tract. Dermal sinus tracts may also cause spinal cord tethering when the attached sinus tract anchors the cord in an abnormal way. Although most of these lesions are located in the lumbosacral area, they can also be found in the cervical and thoracic regions of the spine.

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The incidence of all forms of spinal dysraphism (open and closed) is 0.5 to 8 cases per 1,000 live births. OSD is the most common spinal anomaly. The probability of OSD varies by the clinical presentation and is categorized into low-, intermediate-, and high-risk groups. Children with isolated dimples are in the low-risk group for spinal dysraphism. In children with isolated intergluteal dimples, the probability is 0.34%, and in those with isolated lumbosacral dimples the probability increases to 3.8%. The intermediate-risk group includes children with low or intermediate anorectal malformations, who have a 27% and 33% probability of OSD, respectively. The probability increases to 36% in children with a cutaneous lesion such as a hemangioma, hairy patch, or lipoma. These children are also in the intermediate-risk group. The high-risk group includes children with a high anorectal malformation or cloanal malformation, whose probability for OSD is as high as 44% to 46%. One hundred percent of children with cloacal exstrophy also have OSD. In addition, children with 2 or more congenital midline cutaneous lesions have a 61% to 67% risk of OSD.

OSD is typically asymptomatic in infants and young children, although 50% to 80% of patients have cutaneous lesions on examination. Some individuals remain asymptomatic as adults. However, initial findings in those who develop symptoms are insidious and typically involve autonomic and sphincter dysfunction such as urinary retention, incontinence, urgency or frequency, urinary tract infections, or constipation. Later findings involve sensorimotor symptoms, including leg weakness, decreased or increased muscle tone, and decreased sensation in the legs and perineal area. Left undiagnosed, spinal dysraphism can lead to severe and irreversible neurologic, musculoskeletal, genitourinary, or gastrointestinal disease. Early diagnosis and treatment improves outcomes.

OSD is commonly associated with physical findings on the skin overlying the spine such as a hemangioma, capillary malformation, hair tuft, hypopigmentation or hyperpigmentation, nevus, deviation or duplication of the intergluteal cleft, lipoma, skin tag, or dermal dimple or pit. Of children with OSD, dermal dimples and pits are seen in only 3% to 5% of children with physical findings.

Although dermal dimples or pits may represent a dorsal dermal sinus tract or other cause of OSD, they most commonly are incidental findings in infants and children. Coccygeal or intergluteal dimples and pits occur in 2% to 4% of all children. They are found close to the tip of the coccyx and are usually visible only when the gluteal folds are separated. Although dermal dimples most likely represent benign lesions, they may be the only indication of an underlying spinal dysraphism, and so clinicians must decide what associated features would be an indication for imaging to prevent the less common, but significant, morbidity that can be seen with OSD.

Determining whether to perform imaging studies on infants and children with suspected OSD and selecting the imaging studies are important clinical decisions that many pediatricians face. Any child with neurologic, urologic, or orthopedic signs or symptoms should undergo imaging or be referred to a neurosurgeon. Among infants who do require further evaluation, plain radiographs can reveal vertebral abnormalities but will not detect other causes of OSD and so are generally not recommended. Ultrasonography and magnetic resonance imaging (MRI) are the 2 recommended tests for children with more suspicious lesions. Ultrasonography is not recommended after 3 to 4 months of age because ossification of the vertebral arches makes visualization more difficult.

This decision to image a patient with an abnormal skin finding alone can be informed by knowing the associated probabilities of the abnormal skin finding with OSD. Children with isolated coccygeal or intergluteal dimples or pits do not require imaging because the associated risk of having OSD is 0.34%, whereas those with lumbosacral dimples or pits have a 3.8% risk. Children with lumbosacral dimples (located >2.5 cm from the anus) or with lesions that are greater than 5 mm in size should undergo imaging due to their higher probability (3.8% and 23%, respectively) of OSD than children with isolated coccygeal dimples. The recommended imaging test, ultrasonography or MRI, for infants younger than 3 to 4 months whose only physical sign is a lumbosacral dimple or pit is controversial and often institution dependent. Children older than 3 to 4 months with isolated lumbosacral dimples should be evaluated with MRI because ultrasonography is no longer useful after that age. Children with cutaneous stigma other than an isolated dermal dimple or pit, such as hemangiomas, hair tufts, or lipomas, should be evaluated with MRI at any age because they have a 36% probability of OSD. Children with anorectal malformations, cloanal malformation, or cloacal exstrophy have a 27% to 100% risk of OSD and so should be evaluated with MRI.

Infants with evidence of spinal dysraphism on examination or radiologic testing, as well as children with symptoms suggestive of OSD, should be referred to a pediatric

neurosurgeon for further management. Infants and children with cutaneous lesions along the spine other than a coccygeal dimple or pit can also be referred to a neurosurgeon for further evaluation because these children have a higher risk of spinal dysraphism.

COMMENT: This *In Brief* reminds me of the countless spine examinations I have performed on newborns looking for a dimple or cutaneous lesion, but I am not sure I have always been as vigilant with including this part of the examination with older infants and young children. Infants and young toddlers with OSDs may not have symptoms from urinary or neurologic abnormalities because delay in sphincter control or lower extremity findings may not be readily apparent at their ages. Hence, a careful physical examination of the spine is critical to identify cutaneous physical examination risk factors and patients who warrant further investigation. Concern for OSDs remains the most common indication for spine imaging. Of note, plain radiographs are no longer recommended due to their low specificity of 18% while sensitivity has been found to be 80%. Although there have been no randomized clinical trials evaluating surgery versus medical management in OSD and timing of the surgery, studies to date suggest better outcomes for children who had surgery before 3 years of age. This is an important area where more research is needed to guide our clinical decision making.

– Janet R. Serwint, MD
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ANSWER KEY FOR DECEMBER 2019 PEDIATRICS IN REVIEW

Dysmorphology: 1. D; 2. E; 3. C; 4. D; 5. B.
Congenital Anomalies of the Kidneys, Collecting System, Bladder, and Urethra: 1. E; 2. A; 3. D; 4. B; 5. D.
Ventral Abdominal Wall Defects: 1. A; 2. A; 3. C; 4. E; 5. E.