CUTANEOUS SIGNS OF NEURAL TUBE DYSRAPHISM

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The skin and the nervous system share a common ectodermal origin. Separation of the neural and cutaneous ectoderm occurs early in gestational life, at about the same time that the neural tube is fusing. This embryologic association may explain simultaneous malformations of the skin and the nervous system. The term dysraphia is defined as incomplete fusion of a raphe. This discussion is limited to cutaneous markers of occult neural tube dysraphic conditions in the cranial region (calvarial defects) and those along the spinal axis.

CRANIAL DYSRAPHISM

Cephaloceles

Cephalocele is the general term for congenital herniation of intracranial structures through a scalp defect. Meningoceles are congenital lesions in which only the meninges and cerebrospinal fluid herniate through a calvarial defect. Large encephaloceles and meningoceles pose no diagnostic problem and are easily diagnosed prenatally or at birth. Smaller or atretic encephaloceles and meningoceles may be mistaken for cutaneous lesions, such as hematomas, hemangiomas, aplasia cutis, dermoid cysts, or inclusion cysts. All congenital, exophytic scalp nodules should be evaluated thoroughly; 20% to 37% of congenital, nontraumatic scalp nodules connect to the underlying CNS. Cephaloceles occur in the frontal, parietal, and occipital regions. They are usually midline, although they also may be found 1 cm to 3 cm lateral to the midline. Small cephaloceles are clinically heterogeneous, their appearance dictated by the cutaneous ectoderm.
overlying the lesion. They may be covered with normal skin or have a blue, translucent or glistening surface. A disruption of the surrounding and overlying normal hair pattern is typically present. They are soft, compressible, round or pedunculated nodules that grow when these infants cry or with a Valsalva maneuver.

If a congenital nodule occurs in association with cutaneous abnormalities, the diagnosis of cranial dysraphism is highly suspect. Cutaneous stigmata of cranial dysraphism include hypertrichosis, or the "hair collar sign"; capillary malformations; hemangiomas; and cutaneous dimples and sinuses. The hypertrichosis may overly the nodule, surround a small sinus, or encircle the nodule (i.e., hair collar). A hair collar is a congenital ring of hair that is usually denser, darker, and coarser than the normal scalp hair. When found encircling an exophytic scalp nodule, it is highly suggestive of cranial dysraphism (Fig. 1). The hair collar sign may be found in association with encephaloceles, meningoceles, atretic encephaloceles, atretic meningoceles, and heterotopic brain tissue. A hair collar may also be seen with some lesions of aplasia cutis, so this sign is not entirely specific. Cranial neural tube defects also may be associated with overlying red to pink, blanchable patches that represent capillary malformations. The combination of hair collar sign and capillary malformation surrounding a congenital scalp lesion is almost always indicative of a dysraphic condition. From a clinical standpoint, encephaloceles, meningoceles, atretic cephaloceles, and heterotopic brain tissue are impossible to differentiate, and all should be imaged and surgically corrected by a neurosurgeon as soon as possible to prevent complications.

**Nasal Gliomas**

Gliomas are rests of ectopic neural tissue and differ from frontal encephaloceles in that they do not have a patent intracranial communication. The lesions may be external, intranasal, or combined. Clinically, they are firm, noncompressi-
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Gliomas may be covered with nasal mucosa or normal skin often associated with telangiectasia and misdiagnosed as a hemangioma. They may widen the nasal bone, giving the appearance of hypertelorism. They are congenital and are not progressive, which helps to differentiate them from hemangiomas. Immediate neurosurgical referral is required for surgical removal and reconstruction.

Cranial Dermoid Cyst and Sinus

Dermoid cysts are congenital subcutaneous lesions that are distributed along embryonic fusion lines. The cysts may occur within the fusion lines of the facial processes or within the neural axis. Dermoid cysts result from faulty development and may include dermal and epidermal elements. Although they are always congenital, they may not be noted until early childhood, when they begin to enlarge. They can occur anywhere on the face, scalp, or spinal axis but most commonly are seen overlying the anterior fontanel, at the bregma, the upper lateral region of the forehead, and in the submental region.\(^{25, 27, 28}\) They are nontender, noncompressible, nonpulsatile, cystic blue or skin-colored nodules measuring 1 cm to 4 cm in diameter (Fig. 2). They do not transilluminate or enlarge with a Valsalva maneuver. The overlying skin is normal, unless an external connection exists in the form of a pit or a sinus (Fig. 3).

Midline or nasal dermoid cysts are of greater concern because 25% of these have an intracranial connection.\(^{27}\) Nasal dermoid cysts may occur anywhere from the glabella to the nasal tip; a nasal pit or sinus is present in approximately half of cases (Fig. 3).\(^{27}\) The pit often leads caudally to a dermal sinus and eventuates in a cyst that may be external or within the nasal bones. If the

Figure 2. Nasal dermoid cyst. Firm blue nodule at the glabella giving the appearance of hypertelorism.
dermoid cyst connects to the CNS, cerebrospinal fluid may drain from the sinus. As with nasal gliomas, these patients may have the appearance of hypertelorism if a cyst has widened the nasal bones (see Fig. 2). Nasal dermoids always should be excised because they enlarge over time and damage the nasal bones. Dermoid cysts that are not midline also should be excised because they may develop infection. Radiologic imaging and referral to a neurosurgeon are indicated only for midline dermoid cysts.

Dermal sinuses are 1-mm to 5-mm tracts that connect a dermoid cyst to the skin surface. These usually are midline, found on the nose, occipital scalp, and anywhere along the spinal axis. They may become clinically apparent when they become infected and drain purulent material (Fig. 4). A small tuft of hair is often found protruding from the orifice. If the sinus or cyst directly communicates with the CNS, the patient is at risk for meningitis. The sinus serves as an occult portal of entry for bacteria, often causing a recurrent meningitis that is positive for skin flora. *Staphylococcus aureus* meningitis should be considered secondary to a dermal sinus until proven otherwise. A thorough search for cutaneous fistulas should be carried out and may necessitate shaving the scalp hair. All midline dermal sinuses should undergo radiology before surgical excision. Probing these lesions is contraindicated, given the risk for meningitis.

**SPINAL DYSRAPHISM**

The term *spinal dysraphism* encompasses many congenital anomalies of the spine, including:

- Meningocele
- Myelomeningocele
- Myeloschisis
Occult spina bifida
Diastematomyelia
Diplegemia
Tethered conus
Intraspinal lipoma
Lipomyelomeningocele
Dermoid cyst
Dermal sinus

Larger defects are usually obvious at birth; however, small or occult malformations causing tethering of the spinal cord may be inapparent and asymptomatic at birth. Early diagnosis of a tethered spinal cord is imperative because surgical correction can prevent irreversible neurologic damage. The spinal cord and the spinal canal have the same length in early fetal life. Differential growth leads to progressive ascent of the conus medullaris. At birth, the conus of a healthy infant is at the L3 vertebra and reaches the adult level of L1–L2 at 2 or 3 months of age. If the spinal cord is tethered or bound, it cannot ascend normally and also loses its normal mobility. Flexion of the normal spine causes the cord to move upward; if the cord is fixed or tethered, traction injuries occur and may lead to progressive damage of the lower cord and conus. The tethered cord syndrome is manifest by asymmetric muscle weakness of the lower extremities, scoliosis, back pain, and sensory abnormalities with resultant trophic changes. Approximately 20% of patients have neurogenic bladders and present with repeated urinary tract infections or enuresis. When neurologic symptoms and signs become apparent, most are irreversible, so physicians must diagnose and surgically release the condition before the development of neurologic abnormalities. Overlying cutaneous markers are found in 50% to 90%

Figure 4. Small atrophic patch just right of the midline with a 3 mm nonhealing, crusted erosion. MR image of the spine was normal; however, surgical exploration found a small dermal sinus.
of patients with occult tethered spinal cord. Cutaneous abnormalities may be the only indication of tethered cord, especially in newborn infants.

The cutaneous lesions that should alert physicians to an underlying occult spinal dysraphic state include:

**High index of suspicion**
- Hypertrichosis
- Dimples
- Acrochordons, pseudotails, or true tails
- Lipomas
- Hemangiomas
- Aplasia cutis or scar
- Dermoid cyst or sinus

**Low index of suspicion**
- Telangiectasia
- Capillary malformation (port-wine stain)
- Hyperpigmentation
- Melanocytic nevi
- Teratomas

Most of these lesions are found on or near the midline in the lumbosacral region; however, similar markers in the cervical or thoracic regions also may be indicative of an underlying malformation.

Lumbosacral cutaneous lesions have long been considered markers of spinal dysraphism. The literature suggests that certain skin lesions are more indicative than others are of an underlying malformation. Tavafoghi et al reviewed 200 cases of spinal dysraphism and found that 102 had cutaneous signs. Other studies have documented an even higher prevalence of cutaneous malformations (71–100%). Unfortunately, no prospective studies have been done to determine the percentage of children with cutaneous anomalies in the spinal axis who have occult dysraphism. Interpretation of the literature is difficult because ambiguous terms, such as vascular nevus, telangiectasia, and pigmented macule or nevus, are used.

These cutaneous markers also should be evaluated in the context of a complete history and physical examination, especially in older children. History taking should include questions regarding additional congenital malformations, family history of neural tube defects, weakness or pain in the lower extremities, abnormal gait, scoliosis, difficulties with toilet training or incontinence, recurrent urinary tract infections, and recurrent meningitis. The vertebrae should be palpated for any defects or abnormalities. Examination of the rectum and genitalia is also indicated because associated congenital abnormalities of the urogenital system are often present. The gluteal cleft should be examined carefully for small acrochordons or sinuses; it should be straight and the buttocks, symmetric. A gluteal cleft that deviates is suggestive of an underlying mass, such as a lipoma or meningocele. Examination of the lower extremities is important in older children because they may have trophic changes secondary to nerve damage.

**Hypertrichosis**

Localized lumbosacral hypertrichosis, or “hairy patch,” is usually present in these infants at birth. The hair may be dark or light. The texture of the hair
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Figure 5. Hypertrichosis overlying a tethered cord.

also varies but is frequently described as silky (faun tail nevus). The hypertrichosis is often V-shaped and poorly circumscribed (Fig. 5). Prominent hypertrichosis commonly is associated with other cutaneous stigmata of spinal dysraphism and is highly indicative of a spinal defect. Hypertrichosis in the lumbosacral region may be normal, especially in certain racial groups such as African American, Asian, and Hispanic, and whether additional evaluation is indicated is often difficult to discern. Referral to a neurosurgeon for a more complete neurologic examination may be prudent in these cases.

Lipomas

Lipomas associated with spinal dysraphism are thought to be congenital and are also highly indicative of an underlying defect. Lipomas may lie in the dermis or spinal canal and often penetrate from the dermis through a vertebral defect into the intraspinal space (i.e., lipomyelomeningocele). Intraspinal lipomas are a common cause of the tethered cord. Appropriate radiologic investigation of lumbosacral lipomas must be performed before surgical excision, and a neurosurgeon should be involved because small intraspinal connections may be missed, even with the most sensitive radiologic imaging.

Hemangiomas, Telangiectasia, and Capillary Malformations

Hemangiomas are proliferative vascular tumors that are present in 2.6% of infants. In 1986, Goldberg et al. described five children with large sacral hemangiomas and several other associated abnormalities. Three of the five children had lipomyelomeningoceles. In 1989, Albright et al. reported seven infants with lumbar hemangiomas and tethered spinal cords. Several subsequent reports have supported this association. Hemangiomas associated with spinal dysraphism are usually large (> 4 cm) and overlap the midline. A skin defect or ulceration is often present within the hemangioma (Fig. 6). The hemangiomas
Ulcerated hemangioma overlying a lipomyelomeningocele. Surgery was complicated by wound infection, dehiscence, and cerebrospinal fluid leak.

may be associated with other cutaneous stigmata, such as lipoma, acrochordon, or dermal sinus. These patients are difficult to manage because the hemangiomas can ulcerate, and surgical repair of the tethered cord may have to be delayed until the hemangioma partially regresses. Reports of "telangiectatic patches" are probably describing nascent or partially regressed hemangiomas.

Enjolras et al. reported two patients with cervical spinal dysraphism with overlying capillary malformations (i.e., port-wine stain). Spinal dysraphism associated with a midline, lumbosacral capillary malformation without additional clinical findings is probably uncommon. Two small studies have shown a small prevalence of spinal malformations associated with a solitary capillary malformation of the lumbosacral region. Additional investigation is needed to completely clarify the need for imaging in these infants. A neurosurgical consultation is probably warranted.

Dimples, Aplasia Cutis, and Congenital Scars

Lumbosacral dimples are common but can be a sign of spinal dysraphism. Most infants with sacral dimples that fall within the gluteal crease are healthy. Dimples that are deep, large (> 0.5 cm), fall within the superior portion
or above the gluteal crease (> 2.5 cm from the anal verge), or associated with other cutaneous markers should be radiologically imaged. Deep dimples may be dermal sinuses communicating directly to the spinal canal. These lesions should not be probed.

Aplasia cutis is a congenital absence of skin and occurs most commonly on the scalp. Some forms of aplasia cutis are thought to be secondary to incomplete closure of embryonic fusion lines. Aplasia cutis rarely has been reported in the lumbosacral region and, in that site, may be associated with underlying spinal dysraphism. Scarlike defects also have been described in patients with spinal dysraphism and may be a variant of aplasia cutis. The scarlike regions found in lumbosacral hemangiomas may be a similar phenomenon.

**Acrochordons, Tails, and Pseudotails**

Acrochordons are small, skin-covered, sessile or pedunculated papules or nodules. Histologically they are composed of epidermis and a dermal stalk. A true human tail (i.e., persistent vestigial tail) is rare and is differentiated from a pseudotail and an acrochordon by the presence of a central core of mature fatty tissue, small blood vessels, bundles of muscle fibers, and nerve fibers. A pseudotail is a stumplike structure and is considered a hamartoma composed of fatty tissue and, often, cartilage. Clinically, these lesions are difficult to distinguish clearly, and all have been associated with spinal dysraphism. Radiologic investigation is indicated preoperatively in all cases.

**DIAGNOSIS**

Definitive diagnosis of neural tube dysraphism is made only at surgery. Radiologic imaging is a sensitive screening method and should be undertaken before surgical intervention. Three radiologic modalities are used for the preoperative diagnosis of dysraphism. The most sensitive study is magnetic resonance imaging. Sonography is an excellent, noninvasive alternative in infants less than 6 months of age. The vertebrae are not yet completely ossified in these infants, and sonography is a relatively inexpensive screening tool. If abnormalities are found, magnetic resonance imaging is required preoperatively. CT scanning may delineate bony defects better and may also be necessary, especially in the nasal region. Plain radiography was used extensively in the past but is not sensitive and should not be used for screening. Magnetic resonance imaging has replaced myelograms, also used in the past. It is often useful to speak to the radiologist before ordering the examination because the technology is changing rapidly and varies by institution.

**SUMMARY**

Cutaneous markers have a crucial role in the detection and diagnosis of occult neural tube dysraphism. Recognition of these stigmata and appropriate radiologic imaging decrease the long-term morbidity of this condition.

*References 8, 13, 15, 17, 18, 22, 32, 34, and 36.*
References


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