Can You Identify This Boy's Growing, Darkening, Hairy Lesion?

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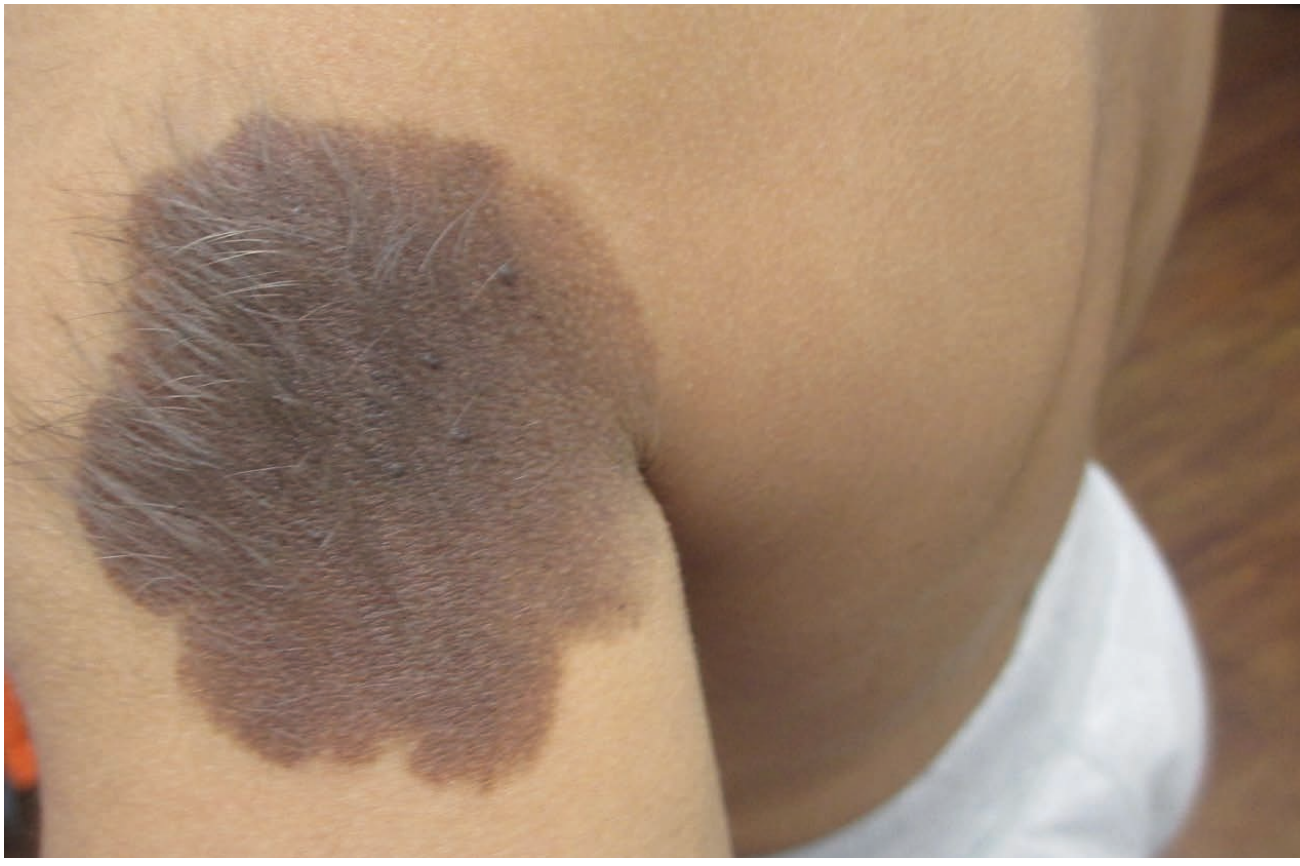
A 3-year-old boy was noted to have a dark, slightly raised skin lesion on his left upper arm at birth. Over time, the lesion became bigger, darker, and hairy. There was no known family history of any similar lesion or melanoma.

Physical examination revealed a 12-cm, discrete, dark plaque on the left upper arm, with small dark papules within it. Tufts of coarse hair were seen over the lesion. There was no other cutaneous or systemic abnormality.

What's your diagnosis?



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Answer: Congenital melanocytic nevus

Congenital melanocytic nevi refer to melanocytic nevi that either are present at birth or develop within the first few weeks of life.[1,2] Persons with congenital melanocytic nevi, mainly the large ones, are at increased risk for melanoma development. Congenital melanocytic nevi pose a variety of diagnostic and therapeutic challenges.

EPIDEMIOLOGY

The incidence ranges from 1% to 2% for any size of congenital melanocytic nevi to approximately 1 in 20,000 to 1 in 500,000 for giant ones.[3-5] Approximately 10% of affected infants have multiple nevi.[2] The female-to-male ratio is approximately 3 to 2.[6] The majority of cases are sporadic, although familial clustering of giant congenital melanocytic nevi has been reported.[7]

PATHOGENESIS

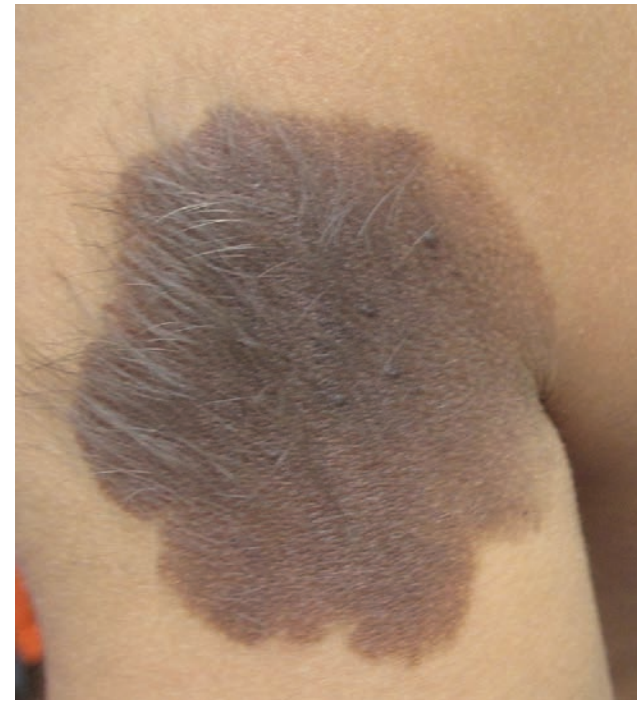
The exact pathogenesis is not known. Presumably, congenital melanocytic nevi arise as a result of disrupted migration of melanoblasts from the neural crest to the skin between the 5th and 24th weeks of gestation.[2,8] It has been shown that neural crest stem cells demonstrate angiotropism or angiocentricity in the embryo.[9] Recently, Kokta and colleagues systematically examined the frequency of angiotropism in 53 cases of congenital melanocytic nevi.[9] Angiotropism associated with microvascular channels was observed in 50 (94.3%) of 53 cases. These findings may explain the origin of neural crest stem cells giving rise to congenital melanocytic nevi.

HISTOPATHOLOGY

Melanocytic nevi are composed of a proliferation of nevus cells that may be present in the dermal-epidermal junction (junctional nevus), in the dermis only (intradermal nevus), or in the junction as well as the dermis (compound nevus). The majority of congenital melanocytic nevi are intradermal or compound in nature, with nevus cells extending more deeply into the dermis than those of acquired melanocytic nevi.[10] The nevus cells can be seen within skin appendages such as hair follicles and sweat glands, between collagen bundles, and surrounding smooth muscles, nerves, and blood vessel walls.[10-12]

CLINICAL MANIFESTATIONS

Congenital melanocytic nevi are classified as being small (< 1.5 cm), medium or intermediate (1.5 cm to 19.9 cm), and large or giant (> 20 cm) according to their expected greatest diameter in adulthood.[4,11,13] Most congenital nevi are small.[2,14] Congenital melanocytic nevi tend to be larger than acquired melanocytic nevi and are usually greater than 5 mm in diameter even at birth.[10]



Congenital melanocytic nevi of the small and medium type are usually round or oval and symmetric.[1,11] Their color varies from light to dark brown. They usually are evenly pigmented.[12] However, more than one color can be present in a congenital melanocytic nevus, and the border of the nevus can be a different hue from the central body of the nevus.[15]

Occasionally, those nevi located on the scalp and face can be pinkish red and, rarely, nonpigmented.[15] The majority of the lesions are palpable but reasonably flat at birth.[10] They may be hairy or hairless.[1] Most nonhairy congenital melanocytic nevi are less than 5 cm in diameter.[6] With time, the lesions tend to become darker and more elevated.[13,16] Coarse dark hair may become prominent in late childhood.[13,16] Congenital melanocytic nevi can involve any location in the skin. Sites of predilection include upper back, chest, lower trunk, shoulders, and proximal limbs.[1]

Giant congenital melanocytic nevi present as dark brown to black plaques, often with a verrucous or cobblestoned surface over time.[6,16] Color variation is common in these lesions, and hypertrichosis often is present. Giant congenital melanocytic nevi frequently are accompanied by multiple smaller, widely disseminated "satellite" nevi.[1,14] These nevi occur most commonly on the posterior trunk and are often known as garment nevi or bathing-trunk nevi.[10]

DIAGNOSIS

Diagnosis usually is a clinical one. Random biopsies of the nevus usually are not helpful. However, biopsy of a newly expanding nodule or suspicious lesion is indicated. Magnetic resonance imaging of the central nervous system (CNS) should

be considered for patients with a giant congenital melanocytic nevus or multiple medium-sized congenital melanocytic nevi, or if CNS melanosis is suspected.[14,17]

DIFFERENTIAL DIAGNOSIS

Congenital melanocytic nevi are distinguished from acquired melanocytic nevi by their larger size, by the lower number of nevi per patient, and by their more varied architecture and a mottled, heterogeneous morphology.[1,6] Dermoscopic features suggestive of congenital melanocytic nevi include reticular network, perifollicular hypopigmentation, and "target globules" (globules contained within empty spaces in the network).[6] Histologically, the melanocytes in congenital melanocytic nevi tend to extend deeper into the dermis and even the subcutaneous tissues compared with the acquired ones.[14]

Other differential diagnoses include dysplastic nevus, melanoma, epidermoid nevus, lentigo, Becker nevus, café au lait spots, plexiform neurofibroma, mongolian spots, nevus of Ota, and nevus of Ito.[17]

COMPLICATIONS

Giant congenital melanocytic nevi may result in cosmetic deformities and psychosocial problems for the child.[1,8,18] As many as 30% of children with giant congenital melanocytic nevi have behavioral changes.[15] This may result in or contribute to family stress. Parental anxiety can be significant because of the potential risk of skin malignancy.[15] Erosions or ulcerations may occur especially in giant congenital melanocytic nevi.[15]

Congenital melanocytic nevi, mainly the larger ones, also predispose a person to the development of melanoma.[19] Red flags for melanoma include color change, irregular border, size increase, surface ulceration, and bleeding.[19] In general, the risk of developing melanoma in congenital melanocytic nevi is proportional to the size of the nevus.[12] The overall lifetime risk of malignant transformation is less than 1% for small and medium-sized congenital melanocytic nevi.[2] Giant congenital melanocytic nevi carry a lifetime risk of melanoma of 2% to 6.3%.[2,20,21] The risk is higher in giant congenital melanocytic nevi that arise on the torso in the bathing-trunk distribution.[22]

In one systematic review of 14 articles that reported on melanoma and giant congenital melanocytic nevi,[21] 52 melanomas occurred in 51 of 2,578 (2%) patients with giant congenital melanocytic nevi. Primary melanomas were mostly cutaneous (82%), and 68% were located on the trunk; 7 melanomas were visceral. Melanoma-associated congenital melanocytic nevi exceeded 40 cm for 74%, and 94% had satellite nevi. On the other hand, a lifetime risk of melanoma in giant congenital melanocytic nevi of approximately 6.3% has been estimated using data from the Danish Birth Registry.[20]

The risk for developing melanoma is highest before puberty in large congenital melanocytic nevi and highest after puberty in small and medium-sized congenital melanocytic nevi.[2,8,22] The presence of numerous satellite nevi near a giant congenital melanocytic nevus in a posterior location has the greatest risk of malignant transformation and neurocutaneous melanocytosis.[14,16,18,22,23] The latter is characterized by the presence of an excessive proliferation of melanocytes within the CNS, including the leptomeninges and the brain parenchyma, in the presence of a giant congenital melanocytic nevus or more than 3 congenital melanocytic nevi.[22] Neurocutaneous melanocytosis affects 5% to 10% of persons with a giant congenital melanocytic nevus.[17]

Rhabdomyosarcoma, liposarcoma, malignant blue nevi, and peripheral nerve sheath tumors occur with increased frequency in patients with giant congenital melanocytic nevi.[6,14,16] Occasionally, congenital melanocytic nevi may be associated with occult spinal dysraphism or tethered spinal cord syndrome.[2]

MANAGEMENT

All patients with congenital melanocytic nevi and their parents should be instructed on sun avoidance and sun protection to reduce the risk of skin cancer.[2,15,16,24] The watch-and-wait approach usually is adopted for small and medium congenital melanocytic nevi.[11]

When deciding whether to excise the lesion, cosmetic and psychosocial issues, the potential for malignant transformation, ease of clinical follow-up, complexity of removal, risk of surgery, and functional outcome must be considered.[24] The management must be individualized for each patient.

When the congenital melanocytic nevus is associated with leptomeningeal melanocytosis, surgical removal of the nevus is indicated. In the absence of leptomeningeal melanosis, prophylactic removal of a large congenital melanocytic nevus is controversial. Some experts recommend prophylactic removal of the lesion *in toto*, with excision into the deep fascia.[25] In spite of this, the absolute risk of malignant melanoma, although low, is still considerably higher than in the general population.[24] Other experts recommend partial-thickness excision, laser treatment, curettage, chemical peels, or dermabrasions with regular follow-up.[24,26] A conservative approach, with serial photography of the nevus and follow-up every 6 months for 5 years and every 12 months thereafter, also has been supported.[4] Authors advocating for the latter approach reason that untreated congenital melanocytic nevi can lighten up spontaneously, that routine surgery cannot eliminate the risk of malignancy, and that early surgery carries increased anesthetic risk.[4] Approximately 17% of these lesions will lighten with age.[4,27] Rarely, congenital melanocytic nevi resolve spontaneously with or without an associated halo or vitiligo.[28-30] ■

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