PHOTO ESSAY

An Atlas of Lumps and Bumps, Part 41: Congenital Melanocytic Nevi

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Congenital Melanocytic Nevi

Congenital melanocytic nevi refer to melanocytic nevi present either at birth or within the first few months of life.[1-4] The incidence ranges from 1% to 2% for small and medium congenital melanocytic nevi to around 1 in 20,000 to 500,000 for large ones.[5-8] Approximately 10% of affected infants have multiple nevi.[4] The female to male ratio is approximately 3:2.[9,10] The majority of cases of congenital melanocytic nevi are sporadic, although familial clustering of large congenital melanocytic nevi has also been reported.[11]

The exact pathogenesis is not known. It is believed that 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.[4,12] Somatic *BRAF*V600E mutations and somatic gain-of-function mutations in *NRAS* have been found in a high proportion of small congenital melanocytic nevi and large congenital melanocytic nevi, respectively.[13] Activating mutations of *BRAF* or *NRAS* may result in over-proliferation of nevus cells, with resulting formation of congenital melanocytic nevi.[10,13,14]

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). Most congenital melanocytic nevi are intradermal or compound in nature with nevus cells extending more deeply into the dermis than those of acquired melanocytic nevi.[1-3]

Congenital melanocytic nevi are arbitrarily classified as small (<1.5 cm), medium or intermediate (1.5 cm to 19.9 cm), and large or giant (> 20 cm), according to the projected size of greatest diameter of the lesion in adulthood.[1-3,7,10,15] The scaling factor used to predict the adult size is based on the anatomical location of the lesion.[10] Congenital melanocytic nevi located on the head, upper limb and trunk, and lower limb are predicted to grow by a factor of 1.7, 2.8, and 3.3, respectively.[10] Most congenital nevi are small, but tend to be larger than acquired melanocytic nevi and are usually > 5 mm in diameter even at birth.[1-4,16]

Small and medium congenital melanocytic nevi are usually round (**Figure 1**) or oval (**Figure 2**) with a smooth surface and uniform color **(Figure 3)**.[1-3,10] Their color varies from tan, brown, to black.

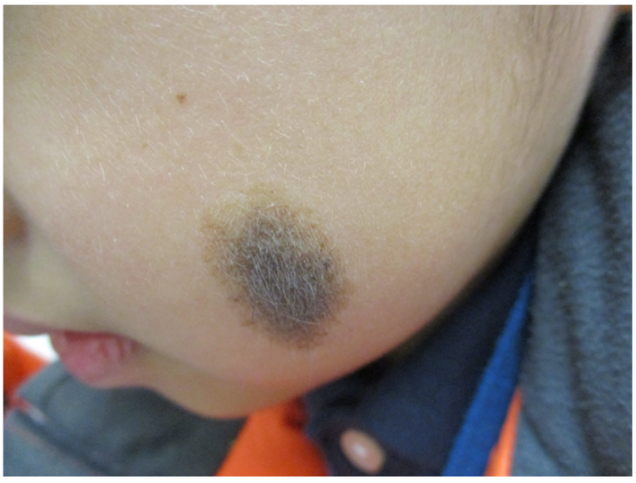

Figure 1. *Small and medium congenital melanocytic nevi are usually round with a smooth surface.*

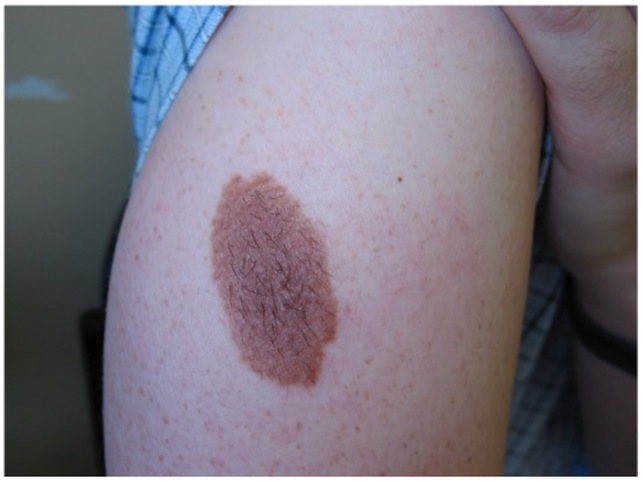

Figure 2. *Small and medium congenital melanocytic nevi are generally uniform in color.*

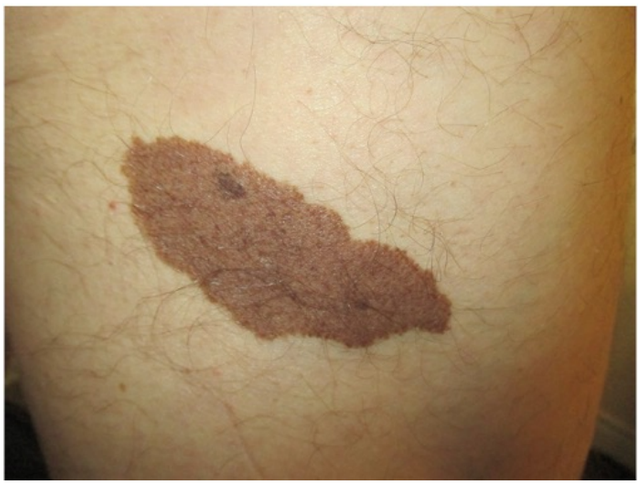

Figure 3. *The color of congenital melanocytic nevi varies from tan, brown, to black.*

They are usually evenly pigmented.[17] 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.[18] Occasionally, those nevi located on the scalp and face can be pinkish red and, rarely, nonpigmented.[18] The majority of the lesions are palpable but reasonably flat at birth.[1,2] They may be hair or hairless.[1,2] Most nonhairy congenital melanocytic nevi are less than 5 cm in diameter.[9] They tend to grow rapidly during early infancy.[13] With time, the lesions tend to become darker and more elevated.[1,2,15] Coarse dark hair may become prominent in late childhood.[1,2,15] Congenital melanocytic nevi can involve any location in the skin. Sites of predilection include upper back, chest, lower trunk, shoulders, and proximal limbs.[1,2] The areola, nipple (Baykal phenomenon) and umbilicus are usually spared.[19,20]

Giant congenital melanocytic nevi present as dark brown to black plaques, often developing a verrucous, cerebriform, mamillated, papillated or cobblestoned surface over time. [1,2,9,14] The borders are often geographic and irregular.[13] Color variegation is common in these lesions and hypertrichosis is often

present. Proliferative nodules may occur.[14] Giant congenital melanocytic nevi are frequently accompanie by multiple smaller, widely disseminated “satellite” nevi. [1,2,16] These giant congenital melanocnevi occu most commonly on the posterior trunk and are often known as garment or bathing trunk nevi.[1,2]

Diagnosis is usually clinical and straightforward, based on the appearance of the lesion and its occurrenc at or shortly after birth. Dermoscopic features suggestive of congenital melanocytic nevi include reticula network, perifollicular hypopigmentation, prominent follicular structures, skin furrow hyperpigmentatio globular pigmentation patterns associated with "target globules" (globules contained within empty space in the network), blotches, and dots.[9,10,13,14] Random biopsies of the nevus are usually not helpful. However, biopsy of a newly expanding nodule or suspicious lesion is indicated.

At times, regression of congenital melanocytic nevi via the halo phenomenon may occur.[3,21,22] When associated with atopic dermatitis that precedes the regression of the nevus, it is known as the Meyerson phenomenon.[23] Regression of congenital melanocytic nevi by sclerosis in the absence of halo phenomenon has also been described.[24]

Giant congenital melanocytic nevi may result in cosmetic deformities **(Figure 4)** and psychosocial problems for the child.[1,2,12,25,26]

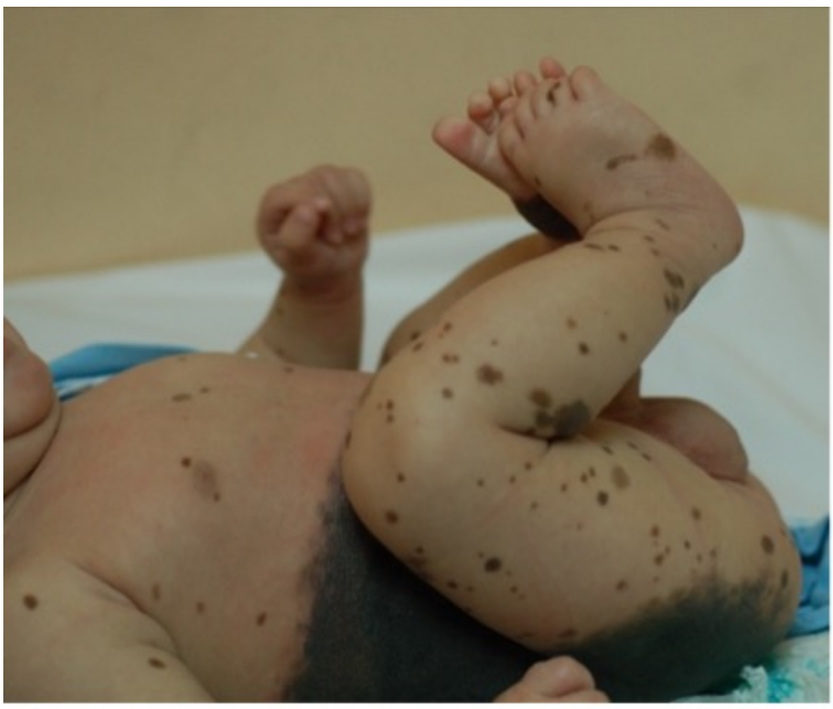

Figure 4. *Giant congenital melanocytic nevi may result in cosmetic deformities*.

As many as 30% of children with giant congenital melanocytic nevi have behavioral changes.[18] This ma result in or contribute to family stress. Parental anxiety can be significant because of the potential risk of skin malignancy.[18] Erosions or ulcerations may occur especially in giant congenital melanocytic nevi.[10,18] **(Figure 5)**

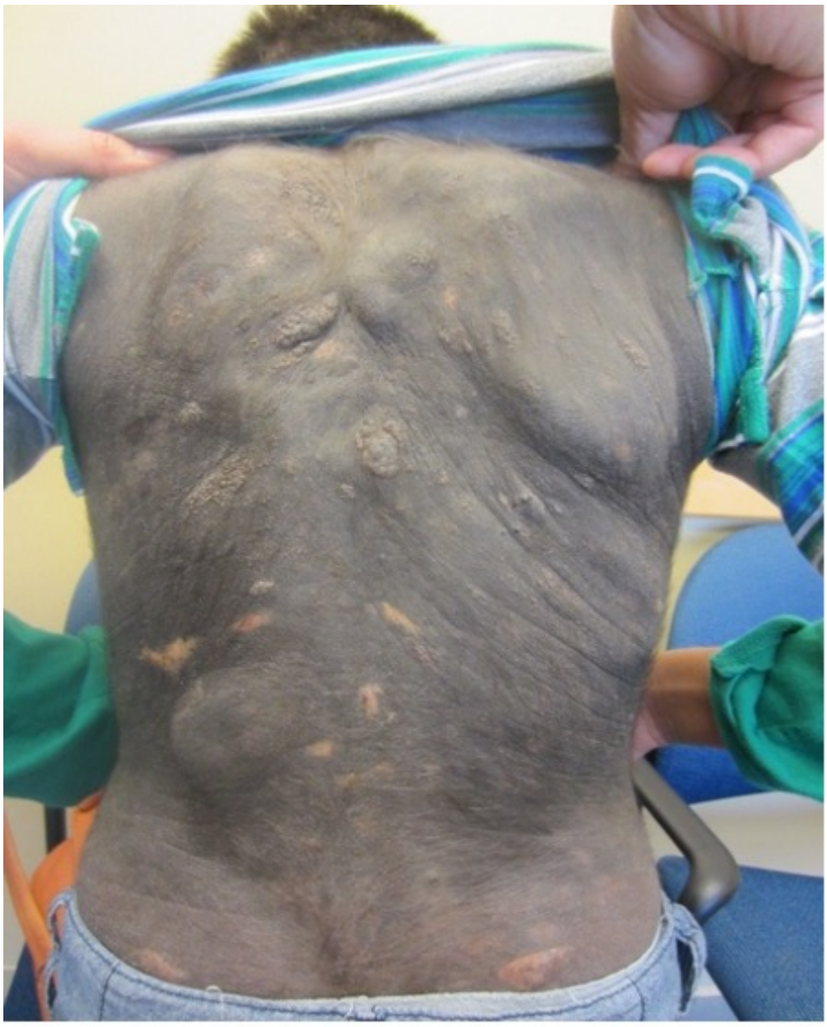

Figure 5. *Giant congenital melanocytic nevi present as dark brown to black plaques, often developing a verrucous, cerebriform, mamillated, papillated or cobblestoned surface over time.*

Congenital melanocytic nevi, mainly the large size ones, also predispose to the development of melanoma.[14,27] Red flags for melanoma include color change, irregular border, increase in size, surface ulceration, and bleeding.[14,27] In general, the risk of developing melanoma in congenital melanocytic nev is proportional to the size of the nevus.[17] The overall lifetime risk of malignant transformation is less tha 1% for small and medium-sized congenital melanocytic nevi.[4] Giant congenital melanocytic nevi carry a lifetime risk of melanoma of 2% to 6.3%.[4] The risk is higher in those giant congenital melanocytic nevi that arise on the torso in the "bathing trunk distribution".[28] 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.[4,12,28] The presence of numerous satellite nevi especially near a giant congenital melanocytic nevus in a posterior location has the greatest risk of malignant transformation an neurocutaneous melanosis.[1,2,16,26,28.29] Neurocutaneous melanosis is characterized by the presence of an excessive proliferation of melanocytes within the central nervous system including the leptomeninges as well as the brain parenchyma in association with a giant congenital melanocytic nevus or more than thre congenital melanocytic nevi.[14,28,30] Neurocutaneous melanosis affects 5 to 10% of patients with a giant congenital melanocytic nevus.[31] Rhabdomyosarcoma, liposarcoma, malignant blue nevi, and malignant peripheral nerve sheath tumors occur with increased frequencies in patients with giant congenital melanocytic nevi.[1,2,9,16] Occasionally, congenital melanocytic nevi may be associated with occult spinal dysraphism or tethered cord syndrome.[4]

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EDITOR'S NOTE:
This article is part of a series describing and differentiating dermatologic lumps and bumps. To access previously published articles in the series, visit: https://www.consultant360.com/resource-center/atlas-lumps-and-bumps.

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