An Atlas of Lumps and Bumps: Part 11

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Pilomatricoma

A pilomatricoma, also known as pilomatrixoma or calcifying epithelioma of Malherbe, is a benign adnexal subcutaneous tumor derived from primitive epidermal germ cells differentiating toward hair matrix cells.[1] Pilomatricomas account for approximately 1% of all benign skin nodules/cysts in childhood.[2] The peak age of onset is in the first 2 decades of life and again between age 50 and 65 years.[1-5] The female to male ratio is approximately 2:1.[2] The condition is more common in White individuals than Asian individuals.[6] Pilomatricomas can be familial.[2] Activating mutations in ß-catenin have been identified in approximately 75% of patients with pilomatricomas.[7] The locus of this tumor has been mapped to the CTNNB1 gene on 3p22-p21.3.[3,7]

Typically, a pilomatricoma presents as a firm to hard, solitary, painless nodule in the subcutaneous tissue **(Figures 1 to 5)**.[1,2] It is usually freely-mobile but slightly attached to the overlying skin.[8] The color of the overlying skin varies from flesh-colored, pink, erythematous, blue, red-blue, to blue-black.[7,9] The size of the lesion is usually 0.5 to 3.0 cm in diameter, although a lesion measuring 34 cm has been reported.[10] Most lesions increase in size slowly over a period of months to years and then stabilize.[1,11] Rapidly growing pilomatricomas have rarely been reported.

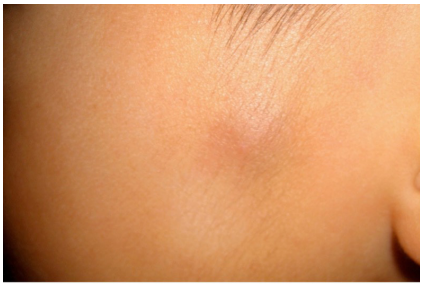

Figure 1. A pilomatricoma presents as a firm to hard, solitary, painless nodule in the subcutaneous tissue.

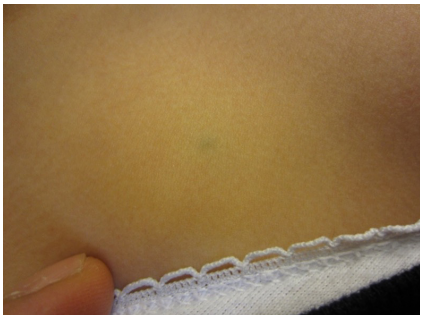

Figure 2. A pilomatricoma is usually freely-mobile but slightly attached to the overlying skin.

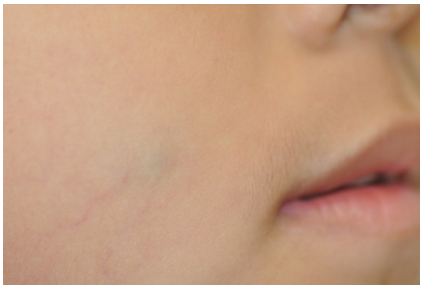

Figure 3. For pilomatricoma, the color of the overlying skin varies from flesh-colored, pink, erythematous, blue, red-blue, to blue-black.

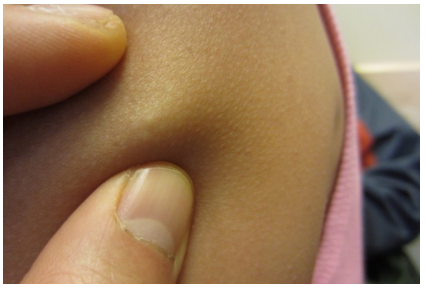

Figure 4. The size of pilomatricoma is usually 0.5 to 3.0 cm in diameter.

The nodule may become hardened if the lesion is calcified. Calcification and ossification occur in 70% to 85% and 15% to 20% of patients, respectively.[1] Downward pressure directed at one end of the lesion may cause the other end to protrude from the skin ("teeter-totter" sign) **(Figure 6)**.[8]

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This article is part of a series describing and differentiating dermatologic lumps and bumps. To access previously published articles in the series, visit https://bit.ly/35J1l1v.

Multiple facets and angles may appear when the overlying skin is stretched ("tent" sign, **Figure 7**).[2,3,8,12] Pilomatricoma most commonly occur on the head (particularly, the face) and neck, followed by upper extremities, trunk, and lower extremities.[2,4,11] The majority of cases are asymptomatic, although some patients may report pain or pruritus.[2]

Several clinical variants have been recognized. In the pseudobullous or anetodermic variant, the lesion is bullous-looking, and the overlying skin is atrophic, translucent, pink, or erythematous **(Figure 8)**.[7] Telangiectasis may be seen. The tumor is rapidly growing. Sites of predilection include the upper arms and shoulders.[13] A pseudobullous or anetodermic pilomatricoma can be depressed at the center when vertical pressure is applied (dimple sign).[13] Rarely, a pilomatricoma may rupture, resulting in an ulcerated or crusted nodule; this variant is referred to as perforating pilomatricoma **(Figure 9)**.[4,7] A pilomatricomal horn is a superficial variant of pilomatricoma.[14] Giant pilomatricoma is another clinical variant, arbitrarily defined as a lesion greater than 5 cm.[7]

Most cases are sporadic. Multiple pilomatricomas occur in 2 to 5% of cases.[1] The presence of 6 or more pilomatricomas is highly suggestive of an underlying disorder such as Gardner syndrome, Turner syndrome, Rubinstein-Taybi syndrome, Kabuki syndrome, Churg-Strauss syndrome, basal cell naevus syndrome (Gorlin syndrome), Soto syndrome, constitutional mismatch repair deficiency (CMMR-D), myotonic dystrophy, xeroderma pigmentosum, sarcoidosis, or trisomy.[7,9,11,12,15-18] Although pilomatricoma is generally benign, malignant transformation has been, very rarely, described.[19,20]

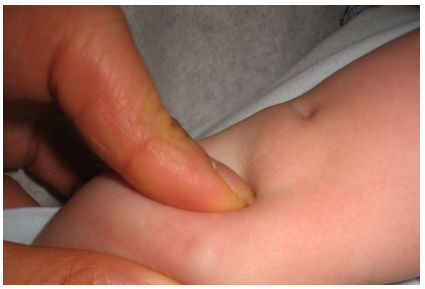

Figure 5. Most pilomatricoma lesions increase in size slowly over a period of months to years and then stabilize.

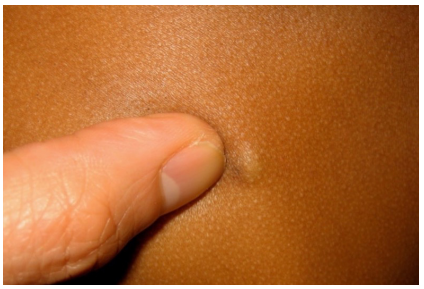

Figure 6. Downward pressure directed at one end of the lesion may cause the other end to protrude from the skin ("teeter-totter" sign).

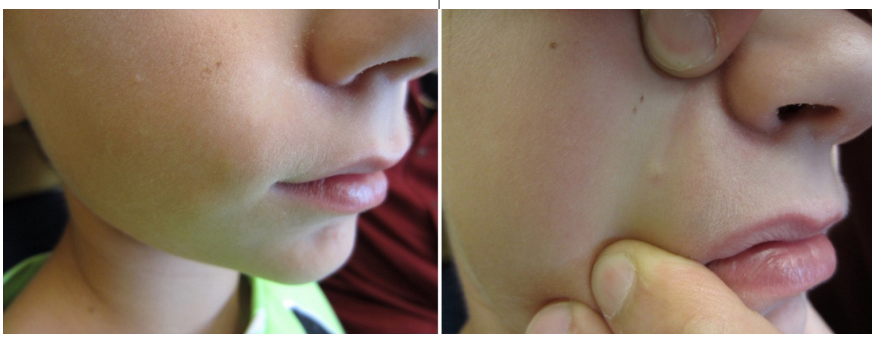

Figure 7. Multiple facets and angles may appear when the overlying skin is stretched ("tent" sign).

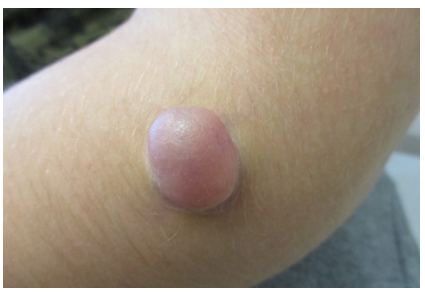

Figure 8. In the pseudobullous or anetodermic variant, the lesion is bullous-looking, and the overlying skin is atrophic, translucent, pink, or erythematous.

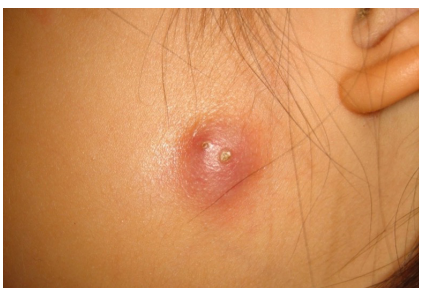

Figure 9. Rarely, a pilomatricoma may rupture, resulting in an ulcerated or crusted nodule; this variant is referred to as perforating pilomatricoma.

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