An Atlas of Lumps and Bumps: Part 11

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 β-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-p21.3,7

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.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.12 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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Multiple facets and angles may appear when the overlying skin is stretched ("tent" sign, Figure 7).\textsuperscript{2,3,8,12} Pilomatricoma most commonly occur on the head (particularly, the face) and neck, followed by upper extremities, trunk, and lower extremities.\textsuperscript{2,4,11} The majority of cases are asymptomatic, although some patients may report pain or pruritus.\textsuperscript{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).\textsuperscript{7} Telangiectasis may be seen. The tumor is rapidly growing. Sites of predilection include the upper arms and shoulders.\textsuperscript{13} A pseudobullous or anetodermic pilomatricoma can be depressed at the center when vertical pressure is applied (dimple sign).\textsuperscript{13} Rarely, a pilomatricoma may rupture, resulting in an ulcerated or crusted nodule; this variant is referred to as perforating pilomatricoma (Figure 9).\textsuperscript{4,7} A pilomatricomal horn is a superficial variant of pilomatricoma.\textsuperscript{14} Giant pilomatricoma is another clinical variant, arbitrarily defined as a lesion greater than 5 cm.\textsuperscript{7}

Most cases are sporadic. Multiple pilomatricomas occur in 2 to 5% of cases.\textsuperscript{1} The presence of 6 or more pilomatricomas is highly suggestive of an underlying disorder such as Gardner syndrome, Turner syndrome, Rubinstein-Taybi 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.\textsuperscript{7,9,12,15-18} Although pilomatricoma is generally benign, malignant transformation has been, very rarely, described.\textsuperscript{2,3,10}

\textbf{References}


