PILOMATRICOMA

Pilomatricoma, also known as pilomatrixoma, is a benign skin lesion thought to be derived from the matrix of hair follicles. It is relatively rare, constituting only 1% of benign skin neoplasms and most commonly present in young children and adolescents. Most pilomatricomas present as a solitary, firm, skin colored papule or nodule located on the head, neck, and upper extremities. There can be a slight discoloration of the area, appearing slightly erythematous or bluish in color. Most lesions are asymptomatic, but can be painful depending on the size and if there is impingement of surrounding structures. Although solitary lesions are most common, multiple pilomatricomas can be seen with certain genetic disorders such as Gardner’s syndrome, myotonic dystrophy, and Rubinstein-Taybi syndrome.

The etiology of pilomatricomas is thought to be due to a somatic mutation in the CTNNB1 gene which regulates the protein beta-catenin. Beta-catenin is a protein involved in cell adhesion and cell signaling and when mutated, leads to uncontrolled cell division and pilomatricoma formation. Most lesions are benign but rarely malignant transformation can occur in middle age to elderly individuals.

A skin biopsy is often performed for confirmation of the diagnosis. The treatment of pilomatricomas is generally surgical removal of the lesions to prevent further growth and disfigurement to the area. Once removed, the lesions are not likely to recur.