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Pilomatricoma

Pilomatricoma, also referred to as pilomatrixoma, is a benign cutaneous neoplasm believed to originate from the hair follicle matrix. Although it is a rare entity, pilomatricoma constitutes approximately 1% of all benign skin tumors. This condition is most commonly observed in children and adolescents, though it can also occur in adults. Pilomatricomas typically manifest as solitary, firm papules or nodules on the head, neck, or upper extremities. Although generally asymptomatic, these lesions may become painful depending on their size and any compression of surrounding structures.

Etiology and Pathogenesis

The development of pilomatricomas is associated with somatic mutations, particularly in the CTNNB1 gene, which encodes the protein beta-catenin. When mutations occur in the CTNNB1 gene, it leads to the accumulation of beta-catenin within the cell, promoting abnormal cell division and the formation of pilomatricomas. Most pilomatricomas are benign, but malignant transformation is rare and typically occurs in older individuals, particularly those in middle age or beyond.

In addition to sporadic occurrences, multiple pilomatricomas may arise in the context of certain genetic syndromes. Notable among these are Gardner's syndrome, myotonic dystrophy, and Rubinstein-Taybi syndrome, where the presence of multiple lesions is often a key diagnostic feature.

Clinical Presentation

Pilomatricomas are typically asymptomatic but can become painful if they compress surrounding structures or if there is associated inflammation. These lesions are most commonly found on the head, neck, and upper extremities, though they may occur anywhere on the body. The lesions often appear as firm, skin-colored or slightly reddish papules or nodules. In some cases, they may present with a bluish hue due to cystic degeneration or secondary inflammation.

Although pilomatricomas are generally solitary, multiple lesions can be seen in patients with underlying genetic conditions such as those previously mentioned. In these cases, the appearance of numerous pilomatricomas warrants further investigation to rule out these syndromes.

Diagnosis

The diagnosis of pilomatricoma is primarily clinical, based on the characteristic presentation of a firm, nodular lesion on the skin. However, for confirmation, a skin biopsy is typically performed.

In cases of multiple pilomatricomas, genetic testing may be recommended to identify mutations associated with conditions like Gardner's syndrome, which is marked by the presence of multiple pilomatricomas along with other characteristic findings, such as osteomas and fibromas.

Management and Treatment

The primary treatment for pilomatricomas is surgical excision. Complete removal of the lesion is generally curative and prevents further growth, as pilomatricomas are unlikely to recur once excised. In cases where the lesion is small and asymptomatic, observation may be appropriate, particularly in pediatric patients. However, if the lesion is symptomatic, large, or located in a cosmetically sensitive area, surgical removal is recommended.

The surgical procedure involves excising the lesion along with any surrounding capsule, ensuring complete removal to reduce the risk of recurrence.

For patients with multiple pilomatricomas associated with genetic syndromes, management may require a multidisciplinary approach, including genetic counseling and regular monitoring for the development of other manifestations associated with these syndromes, such as colorectal polyps in Gardner's syndrome.

Prognosis and Complications

The prognosis for individuals with pilomatricomas is generally excellent, with most lesions being benign and not recurrent after surgical removal. Malignant transformation is rare but can occur in the elderly. Additionally, complications such as infection or scarring can occur, particularly if the lesion is large or located in a sensitive area. Recurrence of pilomatricoma is uncommon but may occur if the excision is incomplete.

Conclusion

Pilomatricoma is a benign skin tumor most commonly affecting children and adolescents. It is characterized by firm, skin-colored nodules, often occurring on the head, neck, and upper extremities. Diagnosis is typically clinical, with confirmation through skin biopsy. The mainstay of treatment is surgical excision, and the prognosis is excellent for most patients. However, awareness of potential genetic associations and the rare risk of malignant transformation is important in managing patients with multiple lesions or those presenting in middle age or older adulthood.

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