Case Report

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Malignant Pilomatrixoma over Left Shoulder: A Case Report

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Pilomatrixoma is a benign neoplasm derived from hair follicle matrix cells. Malignant pilomatrixoma is a rare entity. It is still unclear whether malignant pilomatrixoma arise de novo or it is a malignant transformation in an otherwise benign pilomatrix tumor. 50% of the pilomatrixomas cases occur on the head and neck region, on the cheek, preauricular area, eyelids, forehead, scalp, and lateral and posterior neck. Lesions can also occur on the upper and lower extremities and trunk. Pilomatrixomas are more common in children and are seen more frequently in girls. We present a case of left-arm malignant pilomatrixoma in a 14-year-old girl.

Keywords: Excision, Malignant, Pilomatrixoma, Shoulder

INTRODUCTION

Pilomatrixoma is a benign neoplasm derived from hair follicle matrix cells. It is also referred to as pilomatricoma. It was first identified in 1880 and called as a "calcifying epithelioma." It may be mistaken for a sebaceous cyst, foreign body reaction, soft tissue tumor, or epidermal inclusion cyst. Malignant pilomatrixoma is a rare entity. It is still unclear whether malignant pilomatrixoma arises *de novo* or it's a malignant transformation in an otherwise benign pilomatrix tumor. Approximately, 50% of the lesions occur on the head and neck, especially the forehead, scalp, cheek, eyelids, preauricular area, and lateral and posterior neck.[1] Lesions may also occur on the upper and lower extremities and trunk.[2] Pilomatrixomas are common in children and are found more frequently in girls.[3,4] We present a case of left-arm malignant pilomatrixoma in a 14-year-old girl.

CASE REPORT

A 14-year-old female patient presented to the outpatient department with a history of swelling over the left shoulder since 2 months. The swelling was slow to appear and gradually increased to a size of 2 cm × 2 cm over a period of 2 months. It was painless with no history of trauma.

On examination, it was a 2 cm × 2 cm firm nodule over the left deltoid, non-tender. It was superficial and easily mobile. There was no associated lymph nodal enlargement.

The swelling was excised with aseptic precautions under local anesthesia and primary closure was done. Histopathology revealed nests of large anaplastic hyperchromatic basophilic cells and basaloid cells with scanty cytoplasm and pleomorphic nuclei (Figure 1). Numerous mitotic figures and areas of necrosis were seen. Focal transformation to squamous cells seen was suggestive of malignant pilomatrixoma (Figure 2). A second opinion was taken which confirmed the diagnosis.

During post excision period, patient had a regular follow-up as an outpatient. Patient was asymptomatic, and the wound healed well.

Magnetic resonance imaging (MRI) of soft tissue was performed to rule out any residual tumor (Figure 3). Chest X-ray and ultrasonography abdomen and pelvis was done to rule out any distant metastasis which were negative for metastasis. A wide excision of the scar tissue with 1cm free margins around was done on an elective basis to remove the residual tumor if any (Figure 4). The primary closure of the wound was done.

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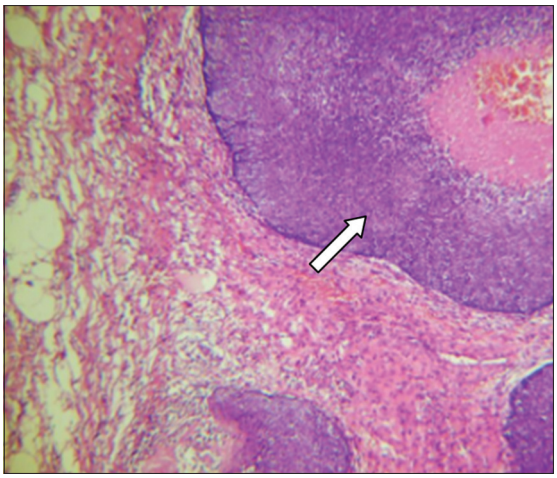

Figure 1: Histopathological picture showing nests of basaloid cells

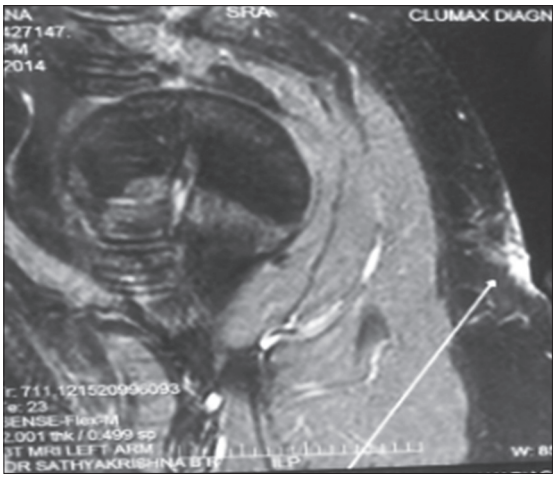

Figure 3: Magnetic resonance imaging picture (after primary excision) showing scar tissue (the report mentioned it as? residual tumor)

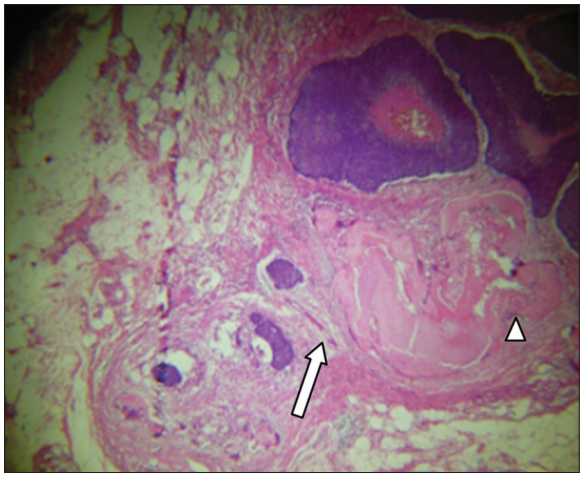

Figure 2: Histopathological picture showing invasion (arrow) to other tissue and ghost cells (arrowhead)

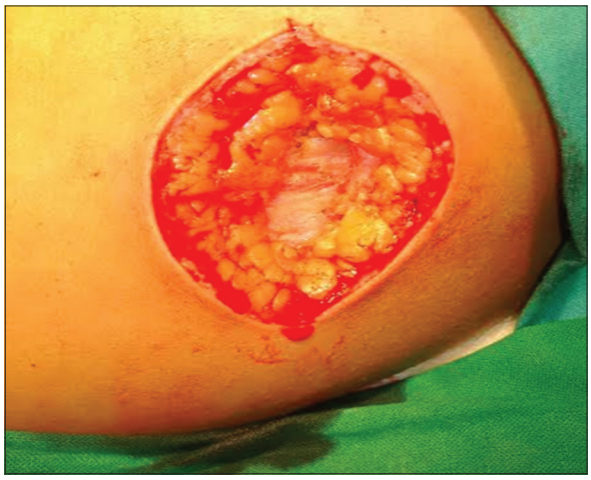

Figure 4: Wide excision of the scar

DISCUSSION

Pilomatrixoma, or calcifying epithelioma of Malherbe, is a benign neoplasm of the skin that originates from hair follicle matrix cells. In 1880, Malherbe and Chenantais were first to describe this lesion, referred to as "calcifying epithelioma," though it was thought to derive from sebaceous glands.[5] The term pilomatrixoma was introduced by Forbis and Helwig in 1961 to better convey the histological origin.[6]

In 1973, Moehlenbeck reviewed 140,000 skin tumors and noted that pilomatrixoma represented 0.12% of cases.[7] It occurs most often in children and young adults. A female preponderance has been seen, with a female: male ratio of 3:2. Furthermore, the majority of patients reviewed in the literature are Caucasian.[7] The lesion primarily affects children and adolescents. Up to 40% of cases occur before the age of 10 and 60% before the age of 20.[8] The highest incidence of this tumor is found in between 8 and 13 years of age.

Pilomatrixomas are usually solitary, but 3.5% of all cases have been reported with multiple foci. Multiple pilomatrixomas and multiple familial pilomatrixomas have been reported to be associated with myotonic dystrophy,[9] Gardner syndrome,[10] and Rubinstein-Taybi syndrome.[11] At least 75% of persons with pilomatrixomas have mutations in the gene CTNNB1; this directly implicates beta-catenin/LEF misregulation as the major causative factor of hair matrix cell tumorigenesis in humans.[12]

Pilomatrixoma develops more frequently in the head and neck region, followed by the upper extremities, the trunk, and the lower extremities.[7] Lesions have been observed in the middle ear and in the ovary.[13,14] Differential diagnosis pilomatrixoma of head and neck region include dermoid cysts, branchial cleft remnants, preauricular sinuses, adenopathy, sebaceous cysts, giant cell tumor, chondroma, foreign body reaction, or malignant soft tissue tumors.[15-17]

Patients usually present with a solitary nodule that exhibits slow growth over several months or years. The diameter

of a pilomatrixoma ranges from 0.5 to 3 cm in majority of cases.[15-17] Lesions are usually the color of the normal skin. Reddish-purple lesions have been observed (probably resulting from hemorrhage). Stretching of the overlying skin can give the lesion a multifaceted, angulated appearance known as the "tent sign," (likely due to calcification in the lesion).[18]

The histopathologic features of a pilomatrixoma include a well-demarcated tumor which is often surrounded by a connective tissue capsule. Generally, it is located in the dermal or subcutaneous layer. The tumor is composed of islands of epithelial cells made up of varying amounts of uniform basaloid matrical cells and often shows the cystic change. Centrally, as the tumor matures, there is degeneration of these basaloid cells. This is characterized by formation of a nucleated ghost (or shadow) cells due to a central unstained areas of these cells.[19] However, these ghost cells, though quite specific, are not unique to pilomatrixomas. There may be a variably prominent inflammatory reaction. Foreign body giant cells, keratin debris, and central calcifications are also characteristic. Calcifications have been seen in 70-85% of cases.[20]

Pilomatrixomas are often misdiagnosed on preoperative evaluation. Fine-needle aspiration cytology has been documented as a preoperative diagnostic method. However, the diagnosis may be misleading without the presence of ghost cells (also known as shadow cells) in the aspirate. Plain X-ray films have limited utility, but may detect calcific foci. The characteristic sonographic picture of pilomatrixoma is an ovoid mass with echogenic center surrounded by a hypoechoic rim with acoustic shadows at the junction of dermis and subcutaneous fat with focal thinning of overlying dermis. It also shows calcification.[21,22] Computed tomography demonstrates a sharply demarcated, subcutaneous lesion of soft tissue density, with or without calcification. MRI may show a rim-enhancing lesion with small areas of signal dropout which may be consistent with calcifications.[23]

Malignant pilomatricoma is a low-grade malignancy, with local evolution and a high recurrence rate in the absence of security margins (up to 50%). Nodal or systemic metastases are uncommon.[24-26] Malignant tumors demonstrate the following features: Poor circumscription, nuclear atypia and atypical mitoses, areas of massive necrosis, infiltration of the hypodermis, dermis or cartilage with sometimes perineural, and/or vascular invasion.[27] Till date, about 80 cases of malignant pilomatrixoma have been reported in the literature.[28] Malignant transformation of pilomatrixoma is rare.[16] Pilomatrix carcinoma typically occurs in the posterior neck or upper back of middle-aged males (male: female ratio of 4:1 in contrast to 2:3[7] for benign lesions). The recurrence rate may be as high as 60% with simple local excision.[29] Rarely, cases of pilomatrix carcinoma with distant metastasis have been reported by Niedermeyer.[26]

The treatment of malignant pilomatrixoma demands aggressive wide local excision with histologically confirmed negative margins. Authors advocate wide excision with a safe margin of 1-2 cm.[25,26] Recurrences after wide excision are rare, with an incidence of 0-3%.[17,23] Radiotherapy could be an alternative. Chemotherapy has not been shown to have a response.[30] Regular follow-up is then mandatory.

CONCLUSION

Malignant pilomatrixoma is an extremely rare malignancy of skin appendages. Diagnosis is complicated and often missed due to shared features with its more common benign counterpart. In patients with recurrence or fast growth of pilomatrixoma, diagnosis of carcinoma should be considered. Wide excision with 1-2 cm margin is the treatment of choice. The role of radiotherapy in advanced cases and recurrent cases has yet to be proved. But, the regular follow-up should be ensured.

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