Cystic Pilomatrixoma: A Diagnostic Challenge
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ABSTRACT

Objectives: 1) Present two childhood cases of preauricular cystic pilomatrixoma; and 2) review the literature on the diagnosis and management of pilomatrixoma.

Study Design: Case report series.

Method: Retrospective review of two childhood cases of cystic pilomatrixoma including a comprehensive review of the relevant literature.

Results: Two children, ages 11 and 7, with cystic preauricular masses are presented. FNA of cyst contents was nondiagnostic and cyst cultures were negative. Computed tomography scans revealed cystic masses superficial to the parotid gland but no distinctive radiologic features characteristic of pilomatrixoma. Complete lesion excision via a preauricular face-lift approach was performed in both cases. The histopathologic features included basoid cells surrounding a cystic space containing keratin debris and characteristic ghost cells consistent with the diagnosis of pilomatrixoma. There has been no evidence of recurrence at 1 and 6 years follow-up, respectively.

Conclusions: Pilomatrixoma is a benign neoplasm which manifests frequently in the head and neck in children, typically with quite classic clinicoradiologic features. However, clinical misdiagnosis may occur in the setting of a preauricular cystic mass presentation. Such cases may be challenging to diagnose even with preoperative imaging and cytology. The treatment of pilomatrixoma is surgical with complete excision yielding a low recurrence rate.

CASE REPORTS

Case 1: A 7-year-old male presented with a right preauricular mass which had gradually increased in size over the previous 6 weeks. The mass was asymptomatic. There had been no antecedent infection or trauma to the area.

Physical exam revealed a 2.5 x 3.5 cm protuberant, red-purple, cystic mass that was minimally tender to palpation (Figure 1). There was significant thinning of the overlying skin with no evidence of fixation. Facial nerve function was intact bilaterally. There was no other masses or lymphadenopathy.

The patient had a white blood cell count (WBC) of 5200, an erythrocyte sedimentation rate of 10mm/hr, and a negative tuberculin skin test (PPD). Computed tomography (CT) and magnetic resonance imaging (MRI) revealed a complex cystic mass that was well-circumscribed which intimately involved the overlying skin. The lesion was superficial to the parotid gland.

After completing a course of amoxillicin-clavulanate without significant change in the appearance of the mass, the patient was taken to the operating room where excision was performed via a preauricular, face-lift approach. The overlying skin was preserved and the mass was removed intact (Figure 2). Aerobic, anaerobic, and mycobacterial cultures were negative. Microscopic examination revealed a cystic lesion lined by basoid cells surrounding a core of keratin debris and ghost cells. These features are diagnostic of pilomatrixoma (Figure 3).

The pilomatrixoma is cystic and surrounded by a dense fibrous stroma with chronic inflammation.

Case 2: An 11-year-old male presented with a left preauricular mass that was noted three weeks prior. The mass was asymptomatic. There had been no antecedent infection or trauma to the area. Of note, the patient had been scratched recently by the family cat.

Physical exam revealed a 2x3 cm cystic mass with reddish discoloration and thinning of the overlying skin. The facial nerve was intact bilaterally and there was no evidence of other masses or lymphadenopathy.

The patient had a WBC of 4900, negative titers for Epstein Barr virus and Bartonella henselae, and a negative PPD. CT revealed a superficial cystic mass overlying the parotid gland (Figure 4). Fine needle aspiration was performed but revealed only dense acute inflammation. Aerobic, anaerobic, mycobacterial, and fungal stains and cultures were all negative. A Warthin Starry stain was negative.

The patient was taken to the operating room for an incision and drainage procedure. Gaseous material from the cyst was sent for pathologic evaluation and microbiologic cultures. The patient was discharged home the following day on a course of amoxicillin-clavulanate. Cultures were again negative; however, pathology revealed abundant ghost cells in sheets, keratin debris, and benign squamous cyst lining cells diagnostic of pilomatrixoma (Figures 5-6).

The patient was discharged home the following day on amoxicillin-clavulanate. Cultures were negative and a PPD revealed no evidence of other masses or lymphadenopathy.

The patient was taken to the operating room one week later at which time the mass was excised intact via a preauricular face-lift approach with preservation of the overlying skin. Pathology confirmed the diagnosis of pilomatrixoma.

DISCUSSION

The classic presentation of pilomatrixoma is a painless, firm, subcutaneous nodule adherent to the overlying skin but mobile relative to underlying structures. There may be bluish or reddish cutaneous discoloration with associated thinning of the epidermis. Growth is slow, typically over months to years, and most masses measure less than 3 cm. The majority of cases occur in the head and neck (50%) and upper extremities (30%), with the lower extremities and trunk (20%) being less common sites. Presentation typically occurs before 10 years of age; cases in adults have, however, been reported. Single lesions are most common (95%); multiple concurrent lesions should raise suspicion for a familial association such as Gardner syndrome.

Although the diagnosis of pilomatrixoma is often suspected in such classic circumstances, alternative diagnoses such as sebaceous cyst and dermoid cyst need to be considered. Clinical features leading to misdiagnosis include atypical location, punctum-like appearance, history of trauma, and cystic consistency.

Cystic pilomatrixomas present a particular diagnostic challenge as the pathognomonic feature of pilomatrixoma— a stony, hard consistency to palpation—is absent. Surprisingly, the cystic variant may represent up to 12% of pilomatrixomas. The differential diagnosis for an inflamed, cystic, cervico-facial mass includes inflammatory lymphadenitis associated with typical bacterial strains as well as atypical mycobacterial infection, branchial remnants, and dermoid/epidermoid cysts. Other entities that may be considered include epidermal inclusion cysts, foreign body reaction and previous trauma.

In a prepuberal child, sebaceous cysts should not be included in the differential diagnosis. Computed tomography (CT) and magnetic resonance imaging (MRI) can help in determining the relationship of a preauricular lesion to the parotid gland, which is useful in distinguishing pilomatrixomas from primary parotid tumors. CT usually shows a non-enhancing, well-demarcated, subcutaneous, opaque lesion, while MRI may demonstrate more soft tissue detail and areas of signal dropout consistent with calcifications. Demonstration of a cystic lesion on CT or MRI, as in our cases, may falsely lead away from the diagnosis of pilomatrixoma.

Fine needle aspirations of pilomatrixomas typically consist of an admixture of basoid epithelial cells, keratinizing squamous cells, anucleate squamous cells with features of ghost cells, calcifications, and multinucleated histiocytes. On gross examination, pilomatrixomas are classically well-circumscribed, grayish-tan masses with a firm to hard consistency. Histopathology reveals lobular islands of basaloid epithelial cells peripherally arranged around cystic spaces containing keratin debris and ghost cells, and lacking a granular cell layer. There is commonly an associated foreign body multinucleated giant cell reaction and fibrosis with calcification, which led to the original description of this lesion as a “calcifying epithelioma of Malherbe.” Pilomatrixoma is near universally a benign lesion. A rare malignant variant, pilomatrix carcinoma, has been reported in adults.

Pilomatrixomas do not spontaneously regress. Progressive growth with overlying skin breakdown and ulceration is possible if left untreated. Surgical excision is the treatment of choice. Although pilomatrixoma histopathologically spares the epidermis, the overlying skin may have to be resected due to fibrosis of tissue planes. If the lesion appears inflamed, the use of antibiotics preoperatively may aid in the dissection allowing skin preservation. Local recurrence is rare unless excision is incomplete.

CONCLUSIONS:

• Pilomatrixoma is a benign neoplasm which manifests frequently in the head and neck in children, typically with quite classic clinicoradiologic features.

• Clinical misdiagnosis may, however, occur, with a preauricular cystic mass presentation; such cases may be challenging to diagnose even with preoperative imaging and cytology.

• The treatment of pilomatrixoma is surgical with complete excision yielding a low recurrence rate.

REFERENCES


