Pilomatrixoma was first described in 1880 by Malherbe and Chenantasi. They described the lesion as a rare, benign subcutaneous tumor arising from the hair cortex. Then in 1961, Forbis and Helwig coined the term “pilomatrixoma” replacing the former identifying name, “calcifying epithelioma of Malherbe.”

Pilomatrixomas usually present as a solitary, slow-growing, subcutaneous mass, and the overlying skin may tend to have a blue-purple discoloration. The majority of cases occur in children under 20 years old. Up to 77% of these lesions are found on the head and neck. The neck is the most common site, followed by the cheek, scalp and periorbital regions. Dermatologists are familiar with the disease, however pilomatrixomas provide a diagnostic challenge to otorhinolaryngologists who do not frequently encounter the presentation. Pilomatrixomas recur without complete surgical excision. Metastasis is rarely found, and mostly documented in case reports.

We present 3 pediatric cases of pilomatrixoma diagnosed on pathology and review the literature for diagnostic challenges related to the diagnosis.

**Methods:** We report 3 cases and review the literature to discuss common factors associated with misdiagnosis and the modalities used.

**Results:** Literature review shows common misdiagnoses are commonly found in lesions with atypical locations, absence of calcifications, and with variable cystic lesions. Misdiagnoses are also found on FNA cytology, and radiologic imaging. CT and MRI imaging are most useful for masses near the parotid to delineate extension of tumor.

**Conclusions:** Pilomatrixoma should be included in the differential when encountering head and neck mass in the pediatric population. Increased awareness of the clinical presentation of the disease will help increase preoperative diagnostic accuracy.

Pilomatrixoma is a preoperative diagnostic challenge. On physical examination, characteristic findings include a bluish discoloration to the skin, and the tendency to be adherent to the skin while mobile over underlying structures. This was seen in one of our case presentations. However, clinicians have found it difficult to diagnose this disease entity accurately preoperatively.

**Literature review shows that preoperative accuracy is achieved only 28-43% cases. Factors associated with this low diagnostic rate include: skin punctum, cystic presentation, absence of clinically appreciable calcification, rapid enlargement, and history of trauma.**

**Patients in our case series contained both cystic presentations and the absence of clinically palpable calcifications.**

Imaging modalities have also contributed to the confusion. Pilomatrixoma characteristically demonstrate a well-circumscribed lesion with homogeneous calcifications on plain radiograph and CT scan. On MRI, these lesions present as a homogeneous mass on unenhanced and gadolinium contrast enhanced studies. However, as seen previously by as our patient who had a CT Neck completed, pilomatrixoma is often missed in the differential. Also previous studies have shown that FNA and PET/CT imaging have misdiagnosed pilomatrixoma for squamous cell carcinoma associated with FDG avid lesions. Due to these findings, radiologic imaging has limited utility in diagnosis and is used mainly for delineating lesions close to the parotid gland.

**References**