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Giant epidermal inclusion cyst masquerading as a soft tissue sarcoma

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ABSTRACT

Epidermal inclusion cysts (EIC) are common lesions formed by the invagination and cystic expansion of the epidermis or hair follicle. We present an unusual case of an epidermal inclusion cyst masquerading as a soft tissue sarcoma based on initial clinical presentation and radiographic findings. Mischaracterization of this type of lesion may lead to unnecessary diagnostic modalities or invasive, overly-aggressive surgery.

1. Background

Epidermal inclusion cysts (EIC) are common lesions formed by the invagination and cystic expansion of the epidermis or hair follicle [1,2]. They are in fact the most common cutaneous cysts and can occur anywhere on the body, but most frequently on the face, scalp, neck, and trunk [1,2]. They are typically skin-colored dermal nodules, often with a central pore or punctum (Fig. 1). Rarely, epidermal inclusion cysts can become inflamed or secondarily infected, causing pain and tenderness, or develop malignancies such as basal cell carcinoma, squamous cell carcinoma, melanoma, or Bowen disease within the cyst [1,2]. Uncomplicated epidermoid cysts are generally a straightforward clinical diagnosis rarely requiring further diagnostic evaluation. We present an unusual case of an epidermal inclusion cyst masquerading as a soft tissue sarcoma. Mischaracterization of this type of lesion may lead to unnecessary diagnostic modalities or invasive, overly-aggressive surgery.

2. Case report

2.1. Presentation (history and physical examination)

A 13-year-old male presented to our pediatric hematology/oncology clinic for evaluation of a slowly growing large left supraclavicular mass. The mass was noticed on a routine health screening examination at the office of his primary care physician the day prior. Ultrasound was obtained at that time, which demonstrated mixed solid and cystic components, most consistent with a preliminary diagnosis of rhabdomyosarcoma. He was subsequently referred to our clinic for further evaluation.

The patient reported that he noticed an expanding mass in his left upper chest about 6 months prior, but did not call attention to it in the hope that it would resolve spontaneously. His father reported that a review of family photographs over the last year demonstrated possible evidence of the lesion through the patient's shirt as far back as 8–9 months prior. The patient stated that the mass was mildly tender to deep palpation, but otherwise not painful and non-pruritic. He denied any other symptoms, such as fever, chills, night sweats, weight loss, shortness of breath, abdominal pain or chest pain. His only known medical problem is mild seasonal allergies; he has no surgical history and denied any recent international travel.

There is a significant family history of diabetes mellitus and hypertension, in addition to a history of unknown “leukemia” in the patient's grandfather, who died at the age of 28, approximately in the 1980s. The patient's maternal aunt also reportedly had a history of multiple small cutaneous hemangiomas. On review of systems, the patient denied any fever, fatigue, unintentional significant weight loss, or other ill symptoms.

On physical examination, the patient was shy and soft-spoken but alert and answering questions appropriately. There was an approximately 8 × 7 × 3 cm, raised, hemispherical, symmetric superficial lesion on the left upper chest wall along the mid-clavicular line (Fig. 2A). Upon visual inspection, the lesion was heterogeneous in appearance, with areas of pallor mixed with hypervascularity and apparent telangiectasias. On palpation, the mass was noted as soft, somewhat mobile, nontender, and non-erythematous. The rest of the physical examination was completely normal, with no telangiectasias, lymphadenopathy, hepatosplenomegaly, bruising, or petechiae.

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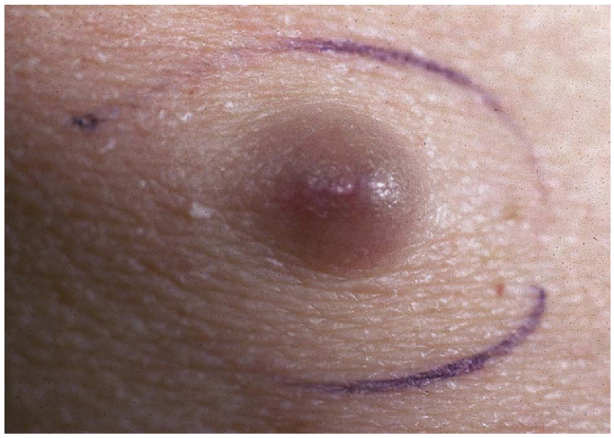

Fig. 1. Typical epidermal inclusion cyst (EIC), located on the back. The lesion is roughly skin-colored, and demonstrates a central punctum. {{86 Burgin, Susan 2017}}.

2.2. Laboratory, imaging, and biopsy results

2.2.1. Laboratory results

Laboratory results were unremarkable, with all electrolytes in the normal range (see Table 1). Inflammatory markers such as C-reactive protein (CRP) and erythrocyte sedimentation rate (ESR) were both in the normal range (less than 5 mg/L, and 4 mm/h, respectively). The heterogenous nature of the patient's mass raised suspicion for a germ cell tumor and so both beta human chorionic gonadotropic (beta-hCG) and alpha fetoprotein (AFP) were measured but were found to be in the normal range (< 2 ng/mL and < 1mIU/mL, respectively). A complete blood count was normal (see Table 1).

2.2.2. Imaging and biopsy

Original ultrasound report from an outside hospital described solid and cystic components most consistent with rhabdomyosarcoma. Repeat ultrasound performed at our facility described a well-defined, heterogenous solid and cystic subcutaneous mass. Doppler imaging showed internal vascular flow in the superior portion of the mass, suggesting a solid component.

CT imaging showed a large well-defined left supraclavicular mass measuring 7.5 × 7.0 × 5.5 cm containing mixed cystic, enhancing solid, and small fatty components, abutting the superior margins of the pectoralis muscle, with no gross invasion of adjacent soft tissue structures and no mass effect (Fig. 2B). The scan was negative for surrounding lymphadenopathy or suspicious lung nodules. The radiographic reading considered a soft tissue sarcoma to be the most likely diagnosis, and thus surgical and pathological consultation was recommended.

Table 1
Laboratory data on presentation.

Comprehensive Metabolic Panel (CMP)	
Sodium	140
Potassium	4.2
Chloride	107
CO2	27
Anion gap	10
BUN	18
Creatinine	0.73
Glucose	89
Total protein	7.4
Albumin	4.2
Calcium	9.6
Total bilirubin	0.4
Alk phosphatase	308 (HIGH)
AST	25
ALT	14
Complete Blood Count (CBC) with differential	
WBC	8.17
RBC	5.41
Hemoglobin	14.4
Hematocrit	43.4
MCV	80.2
MCH	26.6
MCHC	33.2
RDW	13.4
Platelet Count	282
59.1% Neutrophils	
30.6% lymphocytes	
7.7% monocytes	
1.6% eosinophils	
0.6% basophils	

Two days later, the patient underwent ultrasound-guided core needle biopsy under general anesthesia without complication. Initial pathology report was suggestive of epidermal inclusion cyst, but there was concern that this was non-representative given the heterogenous nature of the mass. The patient therefore underwent complete excision of the mass, which is shown in Fig. 3. Histologic examination revealed a mass composed of maturing epidermal lining and keratinaceous debris with an inflammatory response. In areas, the basal layer shows strand like extensions in the surrounding stroma, which was edematous. The latter is a feature of fibrofolliculoma. There is nothing to suggest malignancy. These features provided a final diagnosis of epidermal inclusion cyst with features of fibrofolliculoma.

At the time of publication, the patient is doing well with no recurrence. Post-operative photos of the same area are shown in Fig. 4.

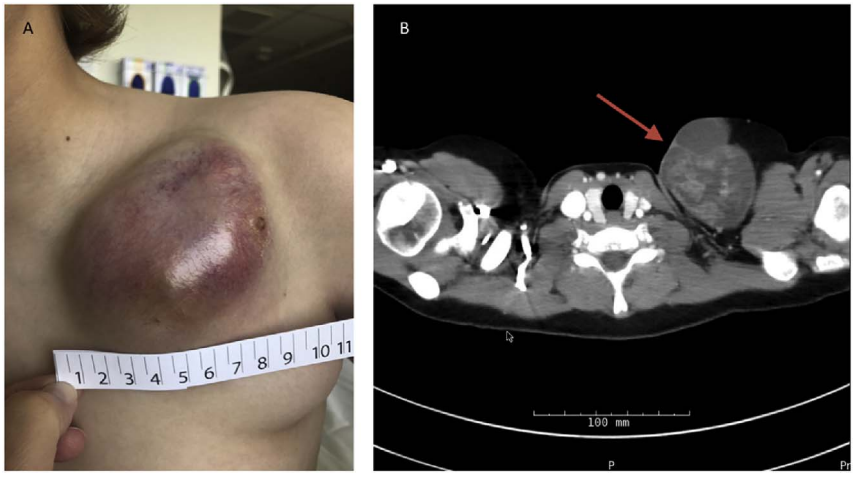

Fig. 2. A: Initial presentation of the patient's chest wall mass. A small, 1 cm punctate lesion can be noted on the left border of the lesion where the patient underwent biopsy. B: The patient's mass as seen on CT, demonstrating a large (7.5 × 7.0 × 5.5 cm) well-defined left supraclavicular mass containing mixed cystic, enhancing solid and small fatty components. The mass abuts the superior margin of the pectoralis muscle, however there is no gross invasion of adjacent soft tissue structures or mass effect. Based on these features, the mass was initially thought to represent a malignant lesion such as a soft tissue sarcoma.