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A subcutaneous manifestation of tuberous sclerosis complex in the posterior scalp

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ABSTRACT

Tuberous sclerosis complex (TSC) is an autosomal dominant syndrome due to a mutation in the TSC2 or TSC1 gene. The disease is known to have variable expressivity involving the neurological, cardiovascular, renal, pulmonary, and integumentary systems (Kennedy et al., 2017). We present a case report, and associated literature review, of a toddler with a posterior scalp lesion which was identified as a soft tissue fibroma upon histopathology. Unlike angiofibromas, soft tissue fibromas in the head and neck are not common in patients with TSC. This soft tissue tumor may be considered as one of the major criteria in the diagnosis of TSC.

1. Introduction

Tuberous sclerosis complex (TSC) is an autosomal dominant syndrome that manifests as cognitive impairment, seizures and hamartomas of the eyes, brain, kidneys, heart, lung and skin [2]. Definitive diagnosis is defined as genetic criterion of mutation in the TSC 1 or TSC 2 gene. However, clinical criteria are still utilized in diagnosis due to the fact that genetic testing may not identify up to 25% of individuals with TSC, and normal gene testing may not exclude TSC [2]. Patients may be considered to have TSC if they present with 2 major criteria or 1 major criterion and 2 minor features. Dental and head and neck specialists should be aware that the presence of three or more angiofibromas or one fibrous cephalic plaque are considered major features. Likewise, minor features include enamel pits and intraoral fibromas [2].

1.1. Case report

A 2 year-old male with a known diagnosis of Tuberous Sclerosis Complex (TSC) presented with a subcutaneous lesion of the posterior scalp measuring approximately 6cm in length by 2cm in height (Fig. 1). Per report, the lesion had been enlarging since birth and began as three separate nodules that had coalesced. Due to the patient's aggressive and self-mutilating behavior, the lesion was constantly becoming traumatized and ulcerated with subsequent bleeding and increased infection risk. Upon examination, the lesion was firm and non-mobile. The overlying skin was largely devoid of hair. There were several areas of increased nodularity within the lesion. A CT of the head (Fig. 2) as well as an MRI were obtained to assess for intracranial extension. After review of risks and benefits of

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surgical removal, the patient's parents elected to proceed with surgical excision of the lesion.

The patient was taken to the operating room and general anesthesia was induced. The patient was positioned in a left lateral decubitus fashion and the proposed incision was marked. Local anesthesia was administered and sufficient time was allowed to elapse for maximum vasoconstrictor effect. The lesion was then removed using a combination of sharp dissection and electrocautery. The lesion was firmly adherent to the overlying dermis in several areas, but was easily dissected in a subgaleal plane. Interestingly, there was deformation of the occipital bone underlying the soft tissue lesion, but no violation of the calvarium. Following removal (Fig. 3), the specimen was sent to surgical pathology for analysis. The excess skin was excised and the incision was closed in layers using 3-0 Vicryl suture to reapproximate the periosteum and galea in an interrupted fashion. The skin was closed using 3-0 chromic suture in a running interlocking fashion. A Glasscock dressing was placed over the wound to prevent the patient from causing direct trauma to the wound.

After specialized staining and testing were carried out, the final surgical pathology report revealed a diagnosis of a tuberous sclerosis associated soft tissue fibroma. The microscopic description indicated that the lesion was formed by bundles of intersecting collagen with scattered spindle cells as well as rounded cells contained within. No giant cells or mitotic figures were seen. Histological features included cytoplasmic reactivity for smooth muscle actin, NKIC3 and factor XIIIa. The infiltrating cells did not stain for S100 protein or melanin-A. The cells were also noted to be focally positive for CD34 (Fig. 4, Fig. 5).

Initially the lesion was thought to be classified as a fibroma-like PEComa which is described as morphologically resembling a fibroma, but with immunochemistry consistent with a perivascular epithelioid cell tumor (PEComa) [3] based on the cytoplasmic reactivity of the specimen for smooth muscle actin and a negative S100 stain. However, given the lack of tumor cells present around blood vessels seen in PEComa this diagnosis was not consistent with the histopathology.

At the time of this publication, the patient has not presented to follow up appointments despite multiple re-scheduled visits.

2. Discussion

Tuberous Sclerosis Complex is a multifaceted disease process. Our patient's degree of disease involvement included cortical dysplasia (tubers), autism spectrum disorder, cardiac rhabdomyomas of the ventricular myocardium, and bilateral simple renal cysts. Neurologically, the patient suffers from epilepsy, aggressive and self-mutilating behaviors as well as intellectual disability.

The common soft tissue lesions of the head and neck, papular angiofibromas and fibrous cephalic plaques, are categorized as major features in the diagnosis of TSC [1,4]. Angiofibromas are viewed as a cosmetic issue but, if large enough, can cause life-threatening hemorrhage, visual impairment, and nasal airway obstruction. The typical facial distribution pattern is along the nasolabial folds, cheeks, and chin. Complete excision of lesions is mandatory as they have a propensity to recur if margins of the specimen are positive [5].

Classically, fibrous tissue tumors are found on the forehead and do not have uniform size, shape, or surface [4]. The posterior scalp location of the lesion in our patient is not typical of TSC, and a literature search failed to return any other reports of lesions in this area

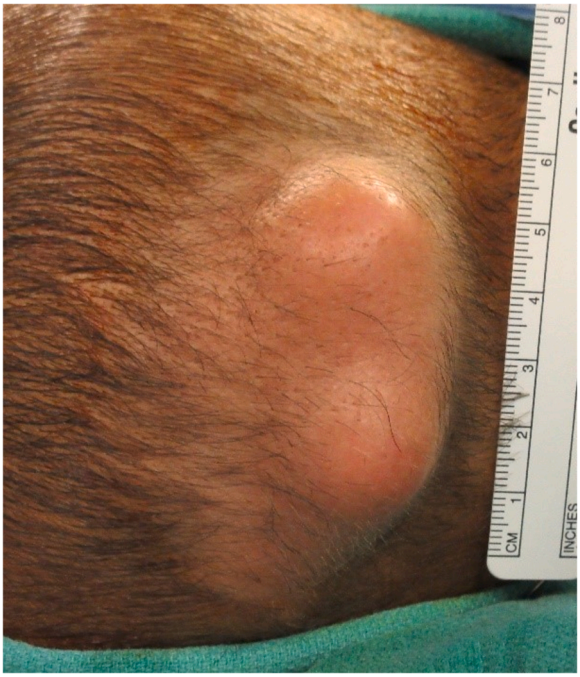

Fig. 1. Soft tissue posterior scalp lesion.

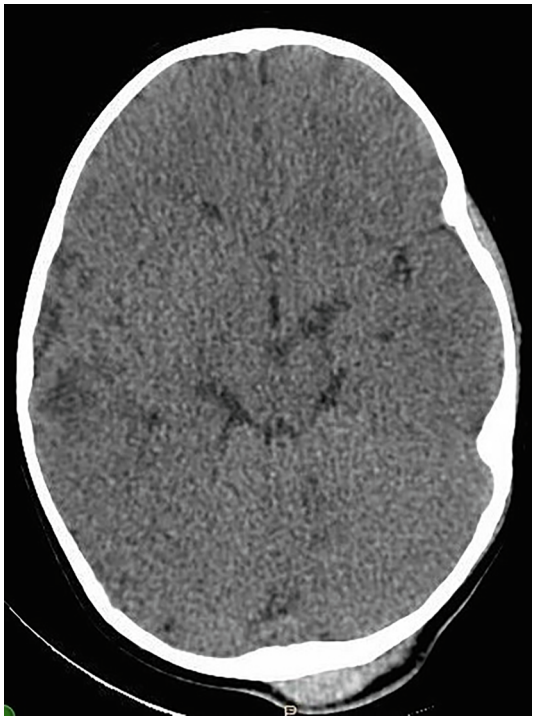

Fig. 2. Pre-operative axial view of a CT Head depicting the subcutaneous lesion of the posterior scalp (soft tissue window).

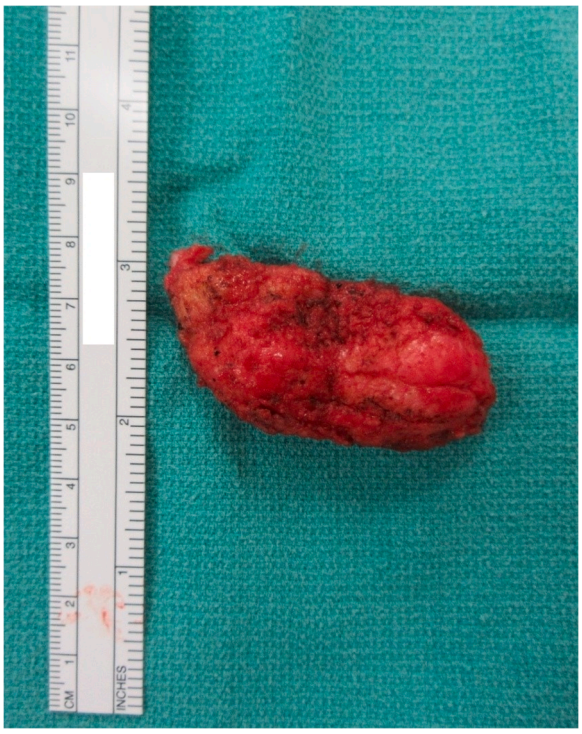

Fig. 3. Excised lesion of the posterior scalp.

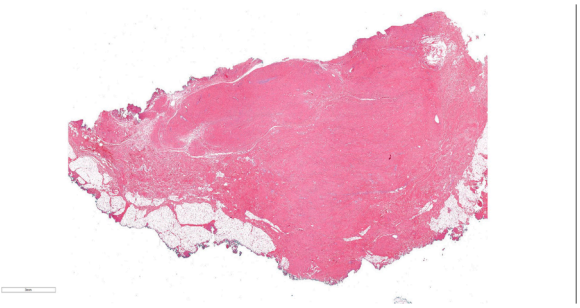

Fig. 4. A densely fibrotic, hypocellular mass replaces the subcutaneous fat and extends into the galea. (Hematoxylin and eosin, 8 X).

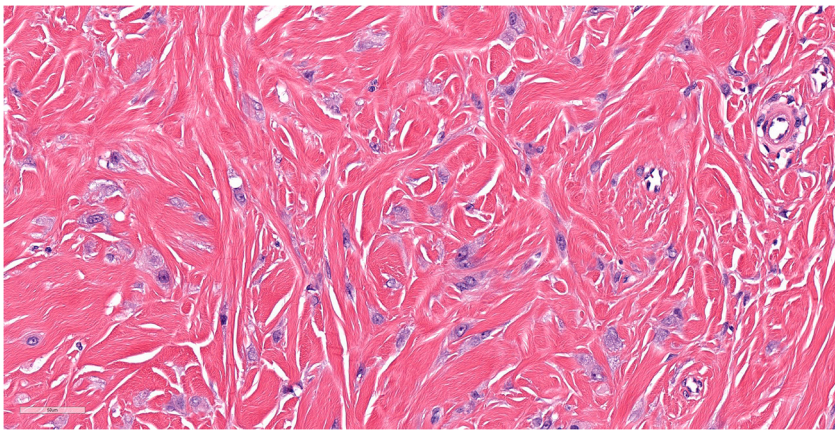

Fig. 5. Scattered bland oval cells are surrounded by dense, hypereosinophilic collagen. No cytological atypia is seen. (Hematoxylin and eosin, 400 X).

of the scalp. According to Baykal et al., fibromatous skin tumors of the scalp were found on 12.6% of their sample size of patients with a diagnosis of TSC. They note all patients with scalp involvement had epilepsy, the most common neurological manifestation of TSC [4]. Treatment of these lesions have ranged from surgical excision, which was performed in our case, to laser therapy as well as use of topical mTOR inhibitors [6–11]. To date, there is a lack of sufficient evidence to generate definitive management recommendations of these lesions [5].

Interestingly, the various lesions that present in TSC have different ages of onset as well as periods of progression, stabilization, and sometimes resolution [2]. For example, angiofibromas begin to appear at age 3, continue to proliferate into the teen years followed by a plateau phase in adulthood [2]. Given that skin lesions may not be apparent from birth or infancy, TSC should not be ruled out based on the absence of skin lesions [2]. With regards to oral manifestations of TSC, many lesions begin in the young childhood years. Hence a baseline oral evaluation is recommended as early as 6 months of age or at the time of diagnosis.

Fibromas of odontogenic origin have been found in association with TSC, presenting as mobility of the dentition and swelling of the gingiva [12]. Intraoral fibromas are considered to be a minor criterion in the diagnosis of TSC. Other common intraoral manifestations include gingival hyperplasia and enamel hypoplasia which is present in nearly 100% of patients [13].

Patients with TSC must have routine surveillance for intraoral lesions, and most patients should undergo oral exams every 6 months. Children with special needs or inability to maintain proper oral hygiene will benefit from more frequent exams. Because intraosseous fibroblastic lesions may present in the maxillofacial skeleton, panoramic radiographic evaluation is recommended by age 6 or if the patient presents with delayed/abnormal tooth eruption [2].

In summary, our patient's presentation of a soft tissue fibroma isolated to the posterior scalp is an uncommon location for a subcutaneous lesion associated with TSC. This lesion is considered one of the major criteria in the diagnosis of the disease. Furthermore, it is important to continue surveillance of soft tissues of the oral cavity, head, and neck following treatment to assess for recurrence.

Conflicts of interest

The authors have no conflicts of interest to disclose.

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