

CASE REPORT

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External auditory canal obstruction due to tuberous sclerosis complex angiofibromas: a case report

Akif İşlek^{1*} and Sadullah Şimşek²

Abstract

Background: Tuberous sclerosis complex (TSC), an autosomal dominant genetic disorder and auricula and external auditory canal (EAC) involvement, is rare. Surgical excision is rarely recommended for cutaneous lesions.

Case presentation: A 31-year-old male patient was admitted with a complaint of left fullness, discharge, and conductive hearing loss. Symmetrical nodular cutaneous lesions on the malar region, tragus, and lateral portion of the EAC bilaterally were noted. The patient had diagnosed with TSC during adolescence clinically and genetically. Surgery was planned to obtain a favorable, self-cleaning EAC rather than total excision of angiofibromas. Angiofibromas on the EAC and tragus were excised. The defect was repaired by sliding a fasciocutaneous island flap inferiorly created in the preauricular area. No EAC stenosis or angiofibroma recurrence was observed during follow-up.

Discussion: Surgical excision for this clinical entity provides satisfactory management even if it was not routinely recommended for other skin lesions.

Keywords: Tuberous sclerosis complex, Neurocutaneous syndromes, Hamartoma, External ear, Island flap

Background

Tuberous sclerosis complex (TSC), an autosomal dominant genetic disorder caused by TSC1 or TSC2 mutations, is characterized by hamartomas in various organs (e.g., skin, brain, lungs, and kidneys) and appears with an incidence rate of approximately 1 in 5000–10,000 [1]. Angiofibromas are the most frequent skin lesions occurred in patients with TSC older than 5 years and characteristically consist of numerous pink to reddish papules or nodules that are typically located on the cheeks, nose, and chin [2]. The traditional and current treatment modalities for facial angiofibromas include topical sirolimus, topical rapamycin cryosurgery, curettage, dermabrasion, and chemical peeling, excision, and

laser therapies. Treatment options generally vary according to the clinical presentation of the disease and the location of the TSC-related lesions. Treatments for skin lesions in the face and head and neck area are usually intended for cosmetic issues, except for ocular manifestations, and the treatment decision is made according to the severity of the disease (e.g., Facial Angiofibroma Severity Index). Auricula and external auditory canal (EAC) involvement in TSC-related angiofibroma is a rare and specific pathology due to the aesthetic and functional feature of the ear.

The obstruction of the EAC causes recurrent infections by preventing aeration and excretion of cerumen. It can also cause conductive hearing loss. Adequate meatoplasty is important to achieve a dry, self-cleaning EAC canal to protect recurrent external otitis. Bony canaloplasty, cartilage excision, Z-plasty techniques, or V-Y flaps are available for EAC meatoplasty [3].

*Correspondence: drakifislek@gmail.com

¹ Acıbadem Eskişehir Hospital, Otolaryngology-Head & Neck Surgery Clinic, Eskişehir, Turkey

Full list of author information is available at the end of the article

Case presentation

The article was structured according to the guideline of CARE [4]. A 31-year-old-male patient was admitted to the Ear Nose Throat Clinic in September 2019 with a complaint of left fullness, discharge, and conductive hearing loss. Symmetrical nodular cutaneous lesions on the malar region, tragus, and lateral portion of the external ear canal (EAC) bilaterally were noted first (Fig. 1). The patient had diagnosed with TSC during adolescence clinically and genetically. The patient also had hypopigmented macules on the trunk and lower extremities and renal angiomyolipoma. There was no neurological symptom and intracranial finding in the patient's previous magnetic resonance imaging (MRI). The patient reported that his father and sibling had similar skin lesions without any neurological symptoms. The complaint about the reason for admission to the Otorhinolaryngology Clinic was recurrent discharge and hearing loss, especially in the left ear. On physical examination, total obstruction of EAC was detected due to TSC angiofibroma (Fig. 2). The lesions were pushed with a rigid endoscope (2.7 mm X 100 mm, Karl Storz SE & Co., Tuttlingen, Germany) by passing through the angiofibromas for EAC examination. On EAC examination, there was a slight purulent



Fig. 1 Multiple facial angiofibromas around the nose



Fig. 2 Total obstruction of external auditory canal due to angiofibromas

secretion and cerumen impaction was detected, and the tympanic membrane was intact. Temporal bone computed tomography showed bilateral soft tissue thickening of the one third external part of EAC. The structures of the middle ear cavity and temporal bone were natural. Bone conduction was normal in the audiometric examination, but the mild conductive hearing loss was detected in the left (pure tone average for 0.5, 1, 2, and 4 kHz; right ear: 15 dB and left ear: 25 dB).

EAC aspiration for cerumen and discharge was performed weekly. Ciprofloxacin/dexamethasone local therapy (Siprogut Plus Drop, 0.3%/0.1%, Bilim Pharmaceutical Co., Istanbul, Turkey) was prescribed for three consecutive weeks. However, no improvement was achieved with medical treatment. So, it was decided to EAC meatoplasty. Surgery was planned to obtain a favorable, self-cleaning EAC rather than total excision of angiofibromas. The surgery was performed under local anesthesia. Angiofibromas on the EAC and tragus were excised. Supraperichondrial dissection was performed especially for the excision of angiofibromas on the external meatus of EAC. The defect was repaired by sliding a 3x2x3 cm fasciocutaneous island flap inferiorly created in the preauricular area (Fig. 3). Skin marking for the required flap was made in the preauricular hairless region according to the size and shape of the defect. Local anesthesia was then infiltrated. The skin incision is complete. To preserve the blood supply to the flap, it was not completely separated from the underlying temporal fascia. Peripheral dissection was performed for adequate movement and rotation. A self-cleaning and well-ventilated EAC was achieved with an open and external meatus.



Fig. 3 External auditory canal patency and adapted preauricular fasciocutaneous island flap

Immunohistochemistry analysis showed that some tumor cells were positive for CD31, actin (SM), CD34 and vimentin, negative expression for smooth muscle actin (SMA), desmin, S100, and AE1/AE3. Ki-67 proliferation index was less than 5%. The pathology specimen was histopathologically diagnosed as angiofibroma. The patient was a positive TSC1 gene mutation. The patient was followed-up with monthly visits for 6 months post-operatively, and no EAC stenosis or angiofibroma recurrence was observed (Fig. 4).

Discussion

TSC is a multisystemic neurocutaneous tumor syndrome caused by mutation of tumor suppressor genes causing hamartomas in different organs such as the skin, brain, lungs, and kidneys [1]. Multiple facial angiofibromas occur in most of the patients with TSC particularly around the nose, appearing as firm skin-colored telangiectatic papules. Also, oral mucosal and gingival fibrous proliferation are another accompanied lesions on the head and neck region [5]. Severe facial and nasal involvement of the disease can cause nasal breathing and feeding difficulty in addition to cosmetic problems. Auricular and EAC skin involvement is an expected but unusual presentation of TSC syndrome. The main complaints were resistant EAC infection and conductive hearing loss in the presented case due to obstruction of EAC.

While topical antineoplastic drugs have priority for common and small lesions on the face, surgical treatment is recommended for more specific lesions and in a limited area due to the risk of scar [1, 2, 6]. Treatments such as dermabrasion, chemical peeling, cryosurgery, and laser surgery are for cosmetic purposes and are insufficient for serious complications such as bleeding and feeding problems [6, 7]. Complete surgical excision of the lesions



Fig. 4 View of the external auditory canal six months after the surgery

is more effective for to solve urgent complications and prevent recurrence [7]. Therefore, surgical excision and reconstruction were considered in the presented case for the treatment of complications due to EAC obstruction. In this case, removing of the lesions on the tragus was aimed to cosmetical results and meatoplasty planned to provide a functional EAC patency. The preauricular fasciocutaneous island flap was adequate for defect reconstruction. The result of the operation was aesthetically and functionally satisfactory for the patient and the surgeon. EAC involvement of TSC is rare and may cause conductive hearing loss and resistant EAC infections by causing a total obstruction. While surgical excision of these lesions provides a permanent solution, it does not cause cosmetic problems in rare involvements such as the auricula.

Conclusion

Auricula and EAC involvement in TSC-related angiofibroma are a rare and specific pathology due to the aesthetic and functional feature of the ear. Surgical excision for this clinical entity provides satisfactory management even if it was not routinely recommended for other skin lesions.

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Authors' contributions

Conceptualization: Aİ and SŞ; Data curation: Aİ and SŞ; Formal analysis: Aİ and SŞ; Investigation: Aİ and SŞ; Methodology: Aİ and SŞ; Project administration: Aİ and SŞ; Resources: Aİ and SŞ; Software: AEK and Aİ; Supervision: Aİ and SŞ; Validation: Aİ and SŞ; Visualization, roles/writing—original draft: Aİ and SŞ; Writing—review and editing: Aİ and SŞ. The authors read and approved the final manuscript.

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929-23/02/2021, Mardin Provincial Health Directorate, Department of Ethics Committees. The patient gave written and verbal consent for the publication.

Consent for publication

A consent form was obtained from the patient.

Competing interests

The authors declare that they have no competing interests.

Author details

¹Acibadem Eskişehir Hospital, Otolaryngology-Head & Neck Surgery Clinic, Eskişehir, Turkey. ²Dicle University, Radiology Clinic, Diyarbakır, Turkey.

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