External Auditory Canal Obstruction due to Tuberous Sclerosis Complex Angiofibromas: Clinical Experience

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Abstract

Introduction Tuberous sclerosis complex (TSC) is characterized by hamartomas or angiofibroma in various organs, and numerous papules or nodules that are typically located on the cheeks, nose, and chin specifically. External auditory canal (EAC) involvement of TSC was not reported and is a challenging issue due to external otitis and conductive hear loss besides aesthetical complaint. This report aimed to examine the effectiveness of surgery for the functional and aesthetic management of the TSC for EAC involvement. Patient and Method A 31-year-old male patient with left auricular mass and ear fullness was presented. The patient's history and family history were compatible with TSC. On physical examination, the total obstruction of EAC was detected due to TSC angiofibroma. Surgical excision and meatoplasty with preauricular fasciocutaneous island flap performed to manage EAC angiofibroma. The pathology specimen was histopathologically diagnosed as angiofibroma. Conclusion Auricula and EAC involvement in TSC related angiofibroma 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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Key Points

1. Angiofibroma of tuberous sclerosis complex (TSC) may occur on the auricula and external auditory canal (EAC).

2. Treatment options vary according to the clinical presentation of the disease.

3. EAC involvement of TSC related angiofibroma a rare and specific pathology.

4. EAC obstruction may cause conductive hearing loss and recurrent external otitis.

5. Excision of the angiofibroma with adequate meatoplasty is important to achieve a dry, self-cleaning EAC.

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

Introduction

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, 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 cosmetical 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 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).

Patient and Method

Preoperative measurements: Article was structured according to the guideline of CARE. A 31-year-old male patient was admitted to the Ear Nose Throat Clinic in Semptember 2019 with complaint of left fullnes, discharge and conductive hearing loss. Symmetrical nodular cutaneous lesions on malar region, tragus and lateral portion of 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 the his father and sibling had similar skin lesions without any neurological symptoms. The complaint for the reason of admission to Otorhinolaryngology Clinic was recurrent discharge and hearing loss, especially in the left ear. On physical examination, total obstruciton 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 secretions and cerumen impaction was detected, the tympanic membrane was intact. Temporal bone Computed Tomography showed bilaterall soft tissue thickening of the one-third externall part of EAC. The structures of middle ear cavity and temporal bone were natural. Bone conduction was normal in audiometric examination, but mild conductive hearing loss was detected in the left (Pure tone Average for 0.5, 1, 2, 4 kHz; right ear: 15 dB and left ear: 25 dB).

Treatment regimen: 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) prescirebed for three consequtive week, 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. Suprapeichondrial 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 and 4). 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 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.

Treatment outcome: 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 patients was positive TSC1gene mutation. The patient was followed-up with monthly visits for six months postoperatively, and no EAC stenosis or angiofibroma recurrence was observed.

Discussion

TSC is a multisystemic neurocutaneous tumor syndrome caused by mutation of tumor suppressor genes causing hamartomas in different organs such as 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-coloured telangiectatic papules. Also, oral mucosal and gingival fibrous proliferation is another accompanied lesions on the head and neck region [4]. 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, 5]. Treatments such as dermabrasion, chemical peeling, cryosurgery, laser surgery are for cosmetic purposes and are insufficient for serious complications such as bleeding and feeding problems [5, 6]. Complete surgical excision of the lesions is more effective for to solve urgent complications and prevent recurrence [6]. Therefore, surgical excision and reconstruction was considered in the presented case for the treatment of complications due to EAC obstruction. In this case, remove of the lesions on the tragus was aimed to cosmetical results and meatoplasty planned to provide a functional EAC patency. Preauricular fasciocutaneous island flap was adequate for defect reconstruction. The result of 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 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 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.

References

[1]. Wataya-Kaneda M, Nagai H, Ohno Y, et al. Safety and Efficacy of the Sirolimus Gel for TSC Patients With Facial Skin Lesions in a Long-Term, Open-Label, Extension, Uncontrolled Clinical Trial. Dermatol Ther (Heidelb). 2020;10(4):635-650. doi:10.1007/s13555-020-00387-7.

[2]. Sadowski K, Kotulska K, Schwartz RA, Jóźwiak S. Systemic effects of treatment with mTOR inhibitors in tuberous sclerosis complex: a comprehensive review. J Eur Acad Dermatol Venereol. 2016;30(4):586-594. doi:10.1111/jdv.13356

[3]. Varadarajan VV, Antonelli PJ. Efficacy of the mini-meatoplasty. Laryngoscope. 2020 May;130(5):1294-1298. doi: 10.1002/lary.28186.

[4]. Kennedy RA, Thavaraj S, Diaz-Cano S. An Overview of Autosomal Dominant Tumour Syndromes with Prominent Features in the Oral and Maxillofacial Region. Head Neck Pathol. 2017;11(3):364-376. doi:10.1007/s12105-017-0778-1

[5]. Lujber L, Burián A. Giant nasal mass causing feeding difficulty in tuberous sclerosis. Otolaryngol Head Neck Surg. 2011;145(3):511-512. doi:10.1177/0194599811410280.

[6]. Earnest L, Byrne P, Califano J. Massive facial angiofibroma in a patient with tuberous sclerosis. Otolaryngol Head Neck Surg. 2003;128(1):151-153. doi:10.1067/mhn.2003.49

Figure Legends

Figure 1 Multiple facial angiofibromas around the nose.

Figure 2 Total obstruction of External auditory canal due to angiofibromas.

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

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