Meatus Acusticus Externus Obstruction due to Tuberous Sclerosis Complex Angiofibromas: A Case Report

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Abstract

Tuberous sclerosis complex (TSC), an autosomal dominant genetic disorder characterized by hamartomas in various organs. Auricula and Meatus Acusticus Externus (MAE) involvement of TSC related angiofibroma is a rare and specific presentation of the disease. A 31-year-old male patient with left auricular mass and ear fullness was presented.

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Running Title: Auricular Tuberous Sclerosis Angiofibromas

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Abstract

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Key Words: Tuberous sclerosis complex, Facial Angiofibroma, External auditory canal, Meatoplasty.

Key Clinical Message

Auricular involvement of TSC related angiofibroma is a rare and specific presentation of the disease and surgical excision may be recommended.

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 Meatus Acusticus Externus (MAE) involvement in TSC related angiofibroma a rare and specific pathology due to the aesthetic and functional feature of the ear.

Case Report

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 bilateral meatus acusticus externus were noted first (Figure 1).The patient's history and family history were competible with TSC. On physical examination, total obstruciton of MAE was detected due to TSC angiofibroma (Figure 2). The lesions were pushed with a 2 mm auto-endoscope by passing through the MAE for external auditory canal (EAC) examination. On EEC 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 exteranl part of EAC. The structures of middle ear cavity and temporal bone were natural. EAC aspiration for cerumen and discharge was performed weekly. Ciprofloxacin / dexamethasone local therapy prescirebed for three consequtive week, But normal self-cleaning EAC has not been achieved for 3 weeks. Upon this, it was decided to meatoplasty. The surgery was performed under local anesthesia. Angiofibromas on the MAE and tragus were excised. The defect was repaired by sliding a 3x2x3 cm fasciocutaneous island flap inferiorly created in the preauricular area (Figure 3). Self-cleaning EAC was achieved with an open and MAE. 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.

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.[3] 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 MAE.

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, 4] 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.[4, 5] Complete surgical excision of the lesions is more effective for to solve urgent complications and prevent recurrence.[5] 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 MAE 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.

Conclusion

MAE 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.

Authors' contributions

Akif İşlek: Design of study, data collection, writing, review of the references, critical review, and final approved version.

Sadullah Şimşek: Design of study, data collection, writing, review of the references, critical review, and final approved version.

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Figure 1 Multiple 2-4 mm facial angiofibromas around the nose.

Figure 2 Total obstruciton of MAE due to angiofibromas.

Figure 3 MAE patency and adapted preauricular fasciocutaneous island flap.