Case Report (Olgu Sunumu)

Effective Treatment of Pediatric Tongue Lymphangiomatosis with Sirolimus: A Case Report

Sirolimus ile Pediatrik Dil Lenfanjiomatozisin Etkin Tedavisi: Bir Vaka Sunumu

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Özet

Lenfanjiomatozis, yaygın lenfanjioma gelişimi ile karakterize edilen nadir bir hastalıktır ve genellikle beş yaş altındaki çocukları etkiler. Lenfanjiomlar baş, boyun, ekstremiteler ve gövdede kistik veya lobüle kitleler olarak ortaya çıkan lenfatik malformasyonlardır. Tedavi seçenekleri klinik gözlem, skleroterapi, radyofrekans ablasyonu, lazer tedavisi ve cerrahiyi içerse de etkinlikleri sınırlı olabilmektedir.

Beş yaşındaki kız hasta, dil büyümesi, kötü ağız kokusu, dilde kanama, çiğneme zorluğu, konuşma ve beslenme bozukluğu şikayetleri ile bölümümüze başvurdu. Fizik muayenesinde dili belirgin şekilde büyümüş, mavi-mor renkli, mikroveziküller ve koyu kabuklarla kaplıydı. Manyetik rezonans görüntüleme (MRG) sonucunda mikrokistik dil lenfanjiomu tanısı konuldu. Lezyonun ameliyat edilemez olması nedeniyle hastaya altı ay boyunca 1,8 mg/m²/gün dozunda sirolimus tedavisi uygulandı. Tedavi sonucunda lezyonların boyutunda belirgin azalma görüldü. Üç ayda bir yapılan takiplerde bir yıl içinde tüm semptomlarda önemli iyileşme saptandı.

Dil lenfanjiomu olan çocuklarda tedavi seçenekleri oldukça sınırlıdır. Bulgularımız göstermiştir ki sirolimus yan etkilerinin yönetilebilmesiyle bu hastalık için umut verici bir tedavi seçeneğidir. Bu nedenle biz pediatrik lenfanjiomatozis vakalarında hasta sonuçlarını etkin bir şekilde iyileştirmek için sirolimus ile hızlı bakım ve tedavi yaklaşımını tavsiye etmekteyiz.

Anahtar kelimeler: Lenfanjiomatosis, dil, sirolimus, çocuk lenfanjiom

Abstract

Lymphangiomatosis, a rare condition characterized by the widespread development of lymphangiomas, often affects children under five years of age. Lymphangiomas, a type of lymphatic malformation, frequently manifest as cysts or lobulated masses in the head, neck, extremities, and trunk. While treatment options include observation, sclerotherapy, radiofrequency ablation, laser therapy, and surgery, the effectiveness can be limited.

A 5-year-old girl was admitted to our department with a history of tongue enlargement, foul breath, bleeding, difficulty chewing, and impaired speech and nutrition. Upon physical examination, her tongue was noticeably enlarged, bluish in color, and covered with microvesicles and dark scabs. A diagnosis of microcystic lymphangioma of the tongue was confirmed through magnetic resonance imaging. Due to the inoperability of the lesion, propranolol 2 mg/kg/day was ordered for 3 months. But no improvement was seen. The patient was treated with sirolimus at a dosage of 1.8 mg/m²/day for six months. This treatment led to a noticeable reduction in lesion size. Subsequent follow-up visits, conducted every three months, revealed a significant improvement in all symptoms within one year.

Treatment options for children with tongue lymphangioma are quite limited. Our findings demonstrate that sirolimus is a promising treatment option for this condition, with manageable side effects. We therefore advocate for prompt and efficient care and treatment with sirolimus in pediatric lymphangiomatosis to improve patient outcomes.

Keywords: Lymphangiomatosis; tongue lymphangioma; sirolimus; pediatric lymphangioma

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INTRODUCTION

The diffuse development of lymphangiomas characterizes Lymphangiomatosis. Lymphangioma is a rare lymphatic malformation (LM), classified by the International Society for the Study of Vascular Anomalies (ISSVA), and diagnosed mostly in children under the age of 5 years as cysts or lobulated masses, typically localized in the head or the neck (approximately 60% of cases), proximal extremities, and trunk. The incidence of LM is estimated to be 1.2–2.8 per 1000 births (1-3). Tongue lymphangioma is a rare benign tumor usually diagnosed in infancy and early childhood. Children with tongue lymphangiomas require a multidisciplinary approach to oral care. Multiple blisters, like a pebbly surface, nodules that appear like a cluster of translucent vesicles on the enlarged dorsal surface of the tongue are seen. These lesions may also be diffuse, involving large portions of the tongue, causing macroglossia (4).

Treatment options for lymphatic malformations are observation, sclerotherapy, radiofrequency ablation, laser treatment, and surgical excision. Since functional and cosmetic features are focused on by patients and families, a personalised therapeutic approach depends on LM size, position, growth rate, and type (5). Surgery and sclerotherapy for LM result in only modest efficacy; rapamycin, also known as sirolimus, represents the current standard care for medical treatment. Sirolimus is active against the mammalian target of rapamycin (mTOR), a serine/threonine protein kinase implicated in the pathogenesis of vascular malformation (6). We report a case of tongue lymphangioma with macroglossia that was treated significantly better with Sirolimus.

CASE REPORT

A five-year-old girl was admitted with enlargement of her tongue, bad mouth odor, bleeding, and difficulty chewing, speaking, and nutrition to the Department of Pediatrics. The patient was referred for further diagnosis to the Pediatric Hematology and Oncology clinic. Her past medical history revealed the swelling of the tongue during the newborn period. She was born to healthy non-consanguineous parents following an uneventful full-term pregnancy. On her physical examination, her tongue was markedly swollen, bluish in appearance, surrounded by a red area in the front of 2/3 part with microvesicles and hematic content, and blackish scabs interfering with feeding and mouth closure without relevant systemic conditions **(Figure 1)**.

There was no mass in the head and neck region. The general examination of the patient does not reveal any abnormalities. The complete blood count and biochemistry tests were within normal limits. The diagnosis of microcystic lymphangioma of the tongue was made with magnetic resonance imaging. Since the surgical intervention was impossible, propranolol 2 mg/kg/day was ordered for 3 months. Despite the patient receiving propranolol treatment, no improvement was seen. Her parents reported the progressive swelling of the tongue and bleeding after nutrition. The patient was treated with sirolimus at a dose of 1.8 mg/m2/day for 6 months. The ongoing treatment of Sirolimus caused a decrease in the size of lesions. Follow-up visits, every 3 months, the improvement of all symptoms after one year **(Figure 2)**.

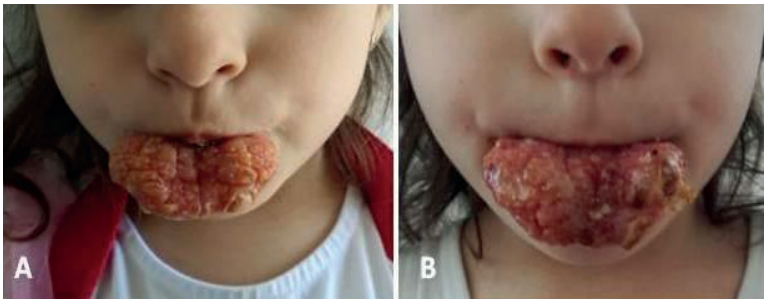

Figure 1. A. Hyperplastic vesicules; B. Brown-yellow nodules

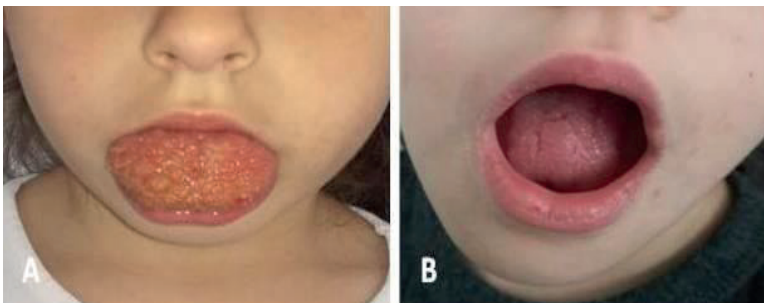

Figure 2. Treatment with Sirolimus: A. At 6 months; B. At one year

DISCUSSION

Lymphangiomas are a relatively rare benign congenital tumor of the lymphatic system (3). When lymphangiomas involve the tongue, a condition frequently referred to as 'macroglossia' can occur and can be associated with a set of problems unique to this anatomic location. Macroglossia may lead to two main types of complications: it can cause poor oral hygiene associated with extensive carious lesions (7,8). In the present case, the patient presented with a history of an enlarged tongue after the age of 5 months. Her tongue continued to swell with a vesicular appearance. Since the diagnosis of microcystic lymphangioma of the tongue was confirmed not until the age of 2 years, she had experienced of feeding and breathing problems.

The clinical appearance of lymphangioma depends essentially on the lesion's extension. Our case presented difficulty in drooling, swallowing, obstruction of the upper airway, and alterations in speech. The superficial localized or diffuse lesions present several translucent vesicles with the same color as that of the adjacent mucosa, or occasionally with a mild reddish blue. Due to infection or hemorrhage, lymphangiomas in a child can grow suddenly but they can also shrink spontaneously (3). There was an enlargement of the tongue progressively in the present case, and due to the risk of potentially fatal complications associated with biopsy, radiologic imaging was emphasized as the primary diagnostic approach. Several studies have demonstrated the utility of WB-MRI in detecting lesions not visible on CT scans and in providing comprehensive anatomic mapping necessary for both diagnosis and follow-up (9).

Treatment modalities of tongue lymphangioma are essentially the restoration of tongue size, the preservation of taste, and the correction of mandibular and dental deformities (7). These congenital malformations can be treated with radiofrequency ablation, surgery, sclerotherapy, interferon, and corticosteroids (8). Since the surgical excision of diffuse microcystic lesions is more complicated, the nonsurgical modalities of treatment may be recommended (10). Although propranolol has proven highly effective for infantile hemangiomas, its efficacy in lymphatic malformations is limited. A case report involving two children aged over 17 months who received oral propranolol at a dose of 2 mg/kg/day demonstrated virtually no reduction in lymphangioma size after 3 months of therapy (11). The present patient was treated with oral Sirolimus, 1.8 mg/kg/day administered in two divided daily doses as first-line treatment with routine follow-up monitoring. Sirolimus can inhibit lymphatic vessel regeneration, invasion and growth. Most recently, it has been reported to be efficacious for patients with lymphangioma of the tongue (12).

Immunosuppressive therapy may cause many acute and long-term side effects, especially in the oral cavity. We checked every 3 months to control the reduction in the size of the tongue. Sirolimus can be effective and safe in patients with complicated vascular abnormalities. The response rates to treatment were excellent and showed significant improvement in signs and symptoms in all children (13). In the present report, a detectable decrease in mass size associated with an evident resolution of the vesicles was observed at 6 months of treatment, and the patient had completely recovered at one year. Any side effects were not recorded during the follow-up.

CONCLUSION

Children with lymphangioma of the tongue, usually presenting with macroglossia, may have significant chewing, speech, and nutrition difficulties. Our findings confirm that sirolimus is a potential treatment for tongue lymphangiomatosis with manageable side effects. For this reason, we recommended that improved care and effective treatment with sirolimus must be done promptly and efficiently in pediatric lymphangiomatosis.

Consent form: An informed consent form was taken from the parent.

Author contribution: The authors declare they contributed equally to the study.

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Conflict of interest: None

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