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Alveoloar Rhabdomyosarcoma of Tongue Base in an Infant : A Case Report

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영아의 설근부에 발생한 폐포성 횡문근육종 1예

충북대학교 의과대학 이비인후과학교실,[1] 해부병리학교실[2]
김지성[1] · 연제엽[1] · 박재홍[1] · 이용문[2] · 이동욱[1]

= 국문초록 =

영아의 설근부에 발생하는 악성 종양은 매우 드물다. 저자들은 연하곤란과 폐쇄성 수면 무호흡증으로 의뢰된 17개월 남아의 설근부에 발생한 횡문근육종을 경험하였기에 문헌고찰과 함께 보고하는 바이다. 외래에서 실시한 이학적 검사 상 설근부 전체가 돌출되어 있었으며 단단한 종괴가 촉지 되었다. 조직검사와 기관절개술을 시행하였으며 컴퓨터단층촬영, 양전자 방출 단층 촬영 그리고 뼈 스캔과 함께 염색체 분석을 실시하였다. 조직검사 결과 폐포성 횡문근육종이 확인되었으며 전이의 증거는 없었다. 염색체 분석상 폐포성 횡문근육종에 상응하는 PAX7-FKHR 유전자 전좌가 발견되었다. 8 회의 항암화학요법과 방사선 치료 후 촉지되는 설근부의 종괴는 없었으며 환자가 호소하는 증상도 개선되었다. 추적 관찰 시 시행된 자기공명영상 결과 확연한 종괴 크기의 감소를 확인할 수 있었다. 횡문근육종은 매우 드문 악성 종양으로 수술과 함께 항암화학요법, 방사선치료 등 여러 치료 방법이 동원되지만 전이나 재발이 있을 시 예후는 매우 불량하다. 그러므로 영아에서 연하곤란, 호흡 곤란 등의 증상이 있을 시에는 설근부를 포함한 상부호흡소화관을 적극적으로 검사하여야 하며 악성 종양의 가능성을 염두에 두어야 하겠다.

중심 단어 : 횡문근육종 · 설근부.

Introduction

Certain processes can cause a focal mass within the tongue base. Focal masses of the tongue base in infants are unusual and can be caused by congenital abnormalities such as thyroglossal duct cyst, lingual thyroid, or vascular or lymphatic malformation. They can also be caused by neoplastic processes such as teratoma or malignancies.[1)] Malignant tumors are extremely uncommon in infants, specifically in the tongue base region.[2)] Rhabdomyosarcoma(RMS) is a highly malignant tumor with local tissue invasion and lymphatic and hematogenous metastasis, and is third in frequency among the solid malignant tumors of childhood.[3)] RMS potentially may arise in any anatomic location, however, the most common site is the head and neck. Despite this predilection, RMS arising in the tongue base is very rare.[4)] Surgery, radiotherapy and chemotherapeutic drugs have been used, alone or in combination, in treating this tumor, however, the outcome for patients with metastatic or recurrent disease remains poor.[5)] Here we report

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a case of RMS of the tongue base in a 17-month-old boy who initially presented with dysphagia and obstructive sleep apnea. We summarize the clinical manifestation, treatments and prognosis of RMS of the tongue base in the literature

Case Report

We present a 17- month-old male infant who suffered from sleep apnea and poor oral intake. His general appearance looked slightly drowsy with arousal difficulty. The infant experienced oral bleeding originating from the tongue base without any trauma history and received primary repair from another hospital 1 month ago.

Physical examination showed a diffuse, firm mass occupying the entire tongue base. Mucosa overlying the mass was intact including the 4 cm suture scar. Computed tomography (CT) revealed a maximum 4.5 cm measured irregular heterogenous enhancing solid and cystic mass at the tongue base with extension to the left sublingual space(Fig. 1).

Tracheostomy was initially done to maintain the airway. Biopsy(frozen section) was proven to be malignant. Due to the potential morbidity from surgical resection, mass excision was not performed.

Permanent tissue biopsy revealed alveolar rhabdomyosarcoma(Fig. 2). Staging workup showed no evidence of metastasis. According to TNM pretreatment staging classification for Intergroup Rhabdomyosarcoma Study IV, patient was stage I(Table 1). Chromosome analysis showed PAX7-FKHR gene translocation which is compatible to alveolar rhabdomyosarcoma.

The patient received 8 cycles of chemotherapy(including vincristine, actinomycin-D, cyclophosphamide) and additional radiation therapy with a dosage of 4,140 cGy. After chemoradiation therapy, there was no palpable mass at the tongue base, patient became symptom free and a 3 month follow up magnetic resonance imaging showed a marked decrease of mass size(Fig. 3).

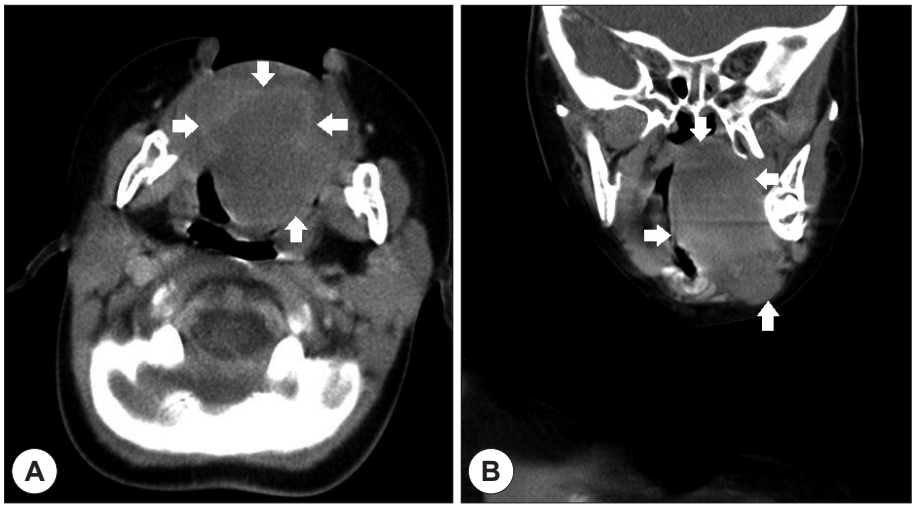

Fig. 1. Enhanced CT images of the mass in axial(A) and coronal(B) cuts. A maximum 4.5 cm irregular heterogenous enhancing solid and cystic mass at tongue base with extension to Lt. sublingual space.

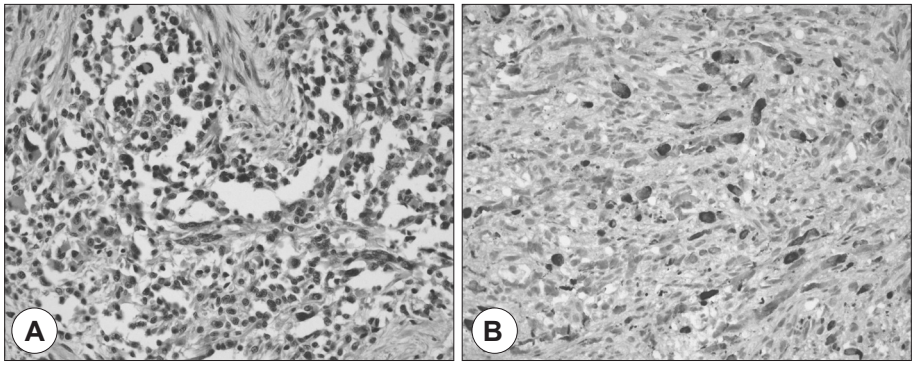

Fig. 2. Nucleus of tumor cells show pleomorphism and mitosis. Fibrous septa separating the lesion and various sized cancer cells with no cohesiveness scattered along the surface of septa, generally mixed multinucleated giant cells are shown. A : Hematoxylin-eosin stain(×400). Multinucleated giant cells stained with desmin which is a differentiation marker between smooth muscle and skeletal muscle cells are shown. B : Desmin stain(×400).

Table 1. TNM pretreatment staging classification for IRS study IV

Stage	Sites	T*	Size	Nodes†	Metastases‡
1	Orbit, head and neck(excluding parameningeal), genitourinary(excluding bladder/prostate)	T1 or T2	A or B	N0, N1, or Nx	M0
2	Bladder/prostate, extremity, cranial parameningeal sites, other (includes trunk, retroperitoneum)	T1 or T2	A	N0 or Nx	M0
3	Bladder/prostate, extremity, cranial parameningeal	T1 or T2	B	N0, N1, or Nx	M0
4	All	T1 or T2	A or B	N0 or Nx	M1

∗ : Tumor : T1, confined to anatomic site of origin ; T2, extension or infiltration into surrounding tissues(A ≤ 5 cm in diameter ; B > 5 cm in diameter), † : Nodes : N0, no distant metastases ; Nx, clinical status unknown ; N1, clinically involved, ‡ : Metastases : M0, no distant metastases ; M1, distant metastases present. IRS : Intergroup Rhabdomyosarcoma Study, TNM : tumor-node-metastases

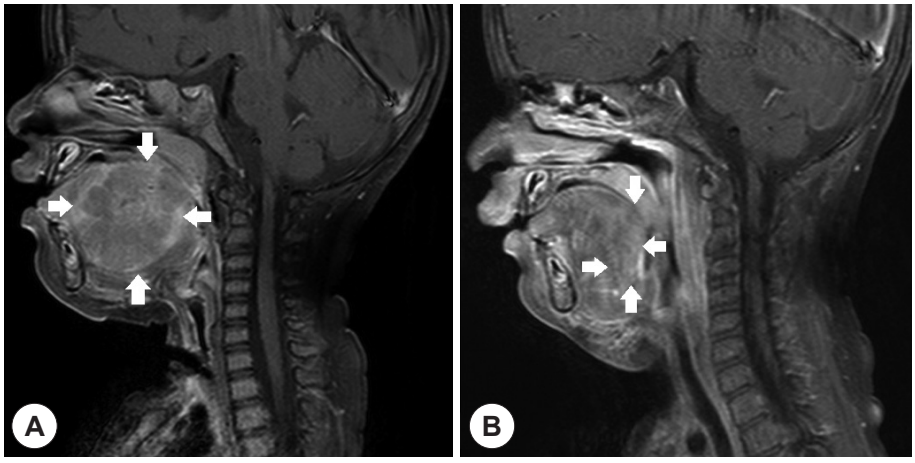

Fig. 3. Pre(A) - and post(B) - treatment enhanced T1- weighted Images. Markedly decreased in extent of enhancing lesion in tong base and sublingual space are shown. The edematous change of subcutaneous fat layer of chin caused by radiation therapy is also noted.

Discussion

Mass in the tongue base can cause lump sensation in the throat, swallowing or breathing difficulty, abnormal phonation, pain if infected, or totally asymptomatic. Patients may also complain of referred otalgia or hemoptysis. Delay in diagnosis is not uncommon because of the vague nature of initial symptoms and the relative inaccessibility of the tongue base examination. Especially in infants, lack of self-expression makes the early detection of tongue base mass difficult. In the case above, detection of the tongue base mass was delayed until dysphagia and obstructive sleep apnea with arousal difficulty had been exacerbated. Thus, symptoms related to swallowing or breathing should raise the suspicion for the upper aerodigestive tract obstruction including mass in the tongue base. Immediate physical examination with intensive history taking, proper imaging or histopathologic studies should be followed, especially in infants. In this case, trauma to the mass from sucking or swallowing may explain the bleeding event of unknown cause in his past medical history.

Being extremely rare in infants, malignant tumor should be included in the differential diagnosis of tongue bass mass and tissue diagnosis should be obtained if the lesion is not proven otherwise. The annual incidence of RMS is about 8/million children. It is the most common soft tissue sarcoma in infants and children and represents about 5 to 15% of all malignant solid tumors.[6] Head and neck RMS occurs most commonly in the orbit, nasopharynx, paranasal sinuses, cheek, neck, middle ear and larynx, however, the occurrence of RMS in the base of tongue is very uncommon.[7]

It is known that RMS arises as a consequence of regulatory disruption of skeletal muscle progenitor cell growth and differentiation[8] and subdivided into three morphological types, alveolar, pleomorphic, and embryonal.[9] Among these, alveolar RMS is highly malignant with a significant incidence of metastatic recurrence and poorly documented to its rarity.[9] Certain genetic conditions are known to increase the risk of RMS, they include, Li-Fraumeni syndrome,[10] Neurofibromatosis type 1,[11] Costello syndrome,[12] Beckwoth-Wiedemann syndrome,[13] and Noonan syndrome.[14] A specific gene translocation, fusion of PAX3 or PAX7 with the FOXO1 gene(ie, FKHR), by the variant t(1 ; 13)(p36 ; q14) is known to exist in the vast majority of cases of alveolar RMS.[15] Within this case, the patient had no involvement with any of the described syndromes, but was proven to be positive for PAX7-

FKHR gene translocation.

Oral and oropharyngeal RMS is commonly treated by radical surgical excision followed by multiagent chemotherapy, usually a combination of vincristine, dactinomycin, and cyclophosphamide. If complete resection is not possible, postoperative radiotherapy may be employed.[16] Recently, improvements in multimodal therapy have resulted in a cure rate of approximately 70% for patients with localized disease[16] and improved the overall five-year survival rate from less than 10% before the 1960s to 65% nowadays.[17] However, about 30 % of patients with localized disease are known to experience recurrence or metastasis during the course of treatment. Metastasis is usually via the bloodstream and less commonly via the lymphatics, usually to cervical lymph nodes, lungs, bones or brain. Five-year survival rate for the patients with recurrence or metastasis remains poor in less than 20%.[5]

In conclusion, RMS is an exceedingly rare, highly malignant tumor. Multi-modality treatments have been used in treating this tumor, but the outcome for patients with metastatic or recurrent disease remains poor. Thus symptoms related to swallowing or breathing in infants such as dysphagia, dysphonia, or obstructive sleep apnea prompt immediate diagnostic work-ups for the upper aerodigestive tract obstruction including the tongue base mass and malignant tumor should be included.

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