International Journal of Pediatric Otorhinolaryngology (2008) 72, 405–408

International Journal of Pediatric Otorhinolaryngology

www.elsevier.com/locate/ijporl

CASE REPORT

Maxillary sinus melanoma as the presenting feature of Carney complex

Matthew W. Ryan [a,*], Samuel Cunningham [b,1], Shu-Yuan Xiao [c,d]

[a] *Department of Otolaryngology, The University of Texas Southwestern Medical Center, 5323 Harry Hines Boulevard, Dallas, TX 75390-9035, United States*
[b] *Quail Creek ENT, Amarillo, TX, United States*
[c] *Department of Pathology, The University of Texas Medical Branch, Galveston, TX, United States*
[d] *Department of Internal Medicine, The University of Texas Medical Branch, Galveston, TX, United States*

Received 4 October 2007; received in revised form 24 October 2007; accepted 26 October 2007
Available online 21 December 2007

KEYWORDS
Mucosal melanoma;
Carney complex;
Maxillary sinus

Summary We present a case of a large maxillary sinus tumor in a 6-year-old boy, immunohistologically indistinguishable from a malignant melanoma, that led to the diagnosis of Carney complex. The Carney complex is an autosomal dominant disorder characterized by mucocutaneous pigmented lesions and neoplasia of multiple endocrine glands and is usually due to an inactivating mutation of the gene for the protein kinase A regulatory subunit 1A. The Carney complex has characteristic head and neck manifestations that can point to the diagnosis of this potentially lethal condition.

1. Introduction

The Carney complex is a multiple neoplasia and lentiginosis syndrome characterized by spotty skin pigmentation, cardiac and other myxomas, endocrine tumors, and other melanocytic tumors. Carney complex is an autosomal dominant disorder with presentation in early childhood and multiple clinical manifestations that develop into adulthood. Some cases are fatal, largely due to cardiac myxomas. Head and neck manifestations, particularly facial skin and mucosal lesions, thyroid nodules and thyroid cancer are prominent components of the complex. Thus, pediatric otorhinolaryngologists should be aware of this rare, but sometimes fatal disease. We report here a case of a child with Carney complex whose presenting feature was a large maxillary sinus tumor.

2. Report of a case

A 6-year-old boy from El Salvador was referred to the Pediatric Otorhinolaryngology clinic at The University of Texas Medical Branch for evaluation of a sinonasal mass. In El Salvador he had been diagnosed with "nasopharyngeal carcinoma of the maxillary sinus" and was partially treated with chemotherapy

* Corresponding author. Tel.: +1 214 648 3637; fax: +1 214 648 9122.
E-mail address: Matthew.ryan@utsouthwestern.edu (M.W. Ryan).
[1] Dr. Cunningham is now in private practice in Amarillo, TX.

0165-5876/$ – see front matter
doi:10.1016/j.ijporl.2007.10.022

before his family moved to the USA. On examination the patient was found to have a maxillary mass causing nasal obstruction, facial and palatal distortion, epiphora, and telecanthus. Computed tomography imaging with and without contrast was obtained (non-contrast image shown in Fig. 1). This study showed a multiloculated expanding lesion involving the maxilla, obliterating the maxillary sinus, and distorting the palate and maxillary dentition. The radiologic appearance was most consistent with an ameloblastoma. Transnasal biopsies of the mass revealed an undifferentiated tumor that was positive for HMB45 and S100 (Fig. 2), consistent with a diagnosis of melanoma. Additionally, at that time a small pedunculated lesion of the eyelid margin was removed and found to be a fibroepithelial polyp—the clinical significance of this small eyelid lesion did not become apparent for years.

The patient underwent total maxillectomy with orbital preservation, and the surgical defect was reconstructed with titanium mesh, a temporalis muscle flap, and obturating dental prosthesis. Surgical margins were clear of disease, and no further adjuvant therapy was used.

The patient was followed over a period of years, with brain and whole body imaging showing no evidence of local recurrence or metastasis. However, he was noted to become overweight and to have evidence of a fatty liver on imaging. He was referred to endocrinology, and 3 years after his initial surgery, hormonal testing confirmed a diagnosis of Cushing's syndrome. Imaging showed adrenal gland enlargement and examination revealed new freckling on his lips and buccal mucosa. The patient underwent bilateral adrenalectomy with pathology showing primary pigmented adrenocortical hyperplasia. At this time, Carney complex was suspected and further screening studies showed a calcified mass of the testis that on orchiectomy was noted to be a Leydig cell tumor. The combination of primary pigmented adrenocortical hyperplasia, facial spotty pigmentation, eyelid skin lesion, and testicular calcification satisfy the diagnostic criteria for the Carney complex. To date, no other Carney complex conditions have been identified, and the patient is receiving close clinical follow-up including routine thyroid ultrasonography. Questioning of the family did not reveal any other family members affected with the syndrome, though the child's father was noted to have freckling on his lips. To our knowledge none of the child's relatives has undergone genetic testing.

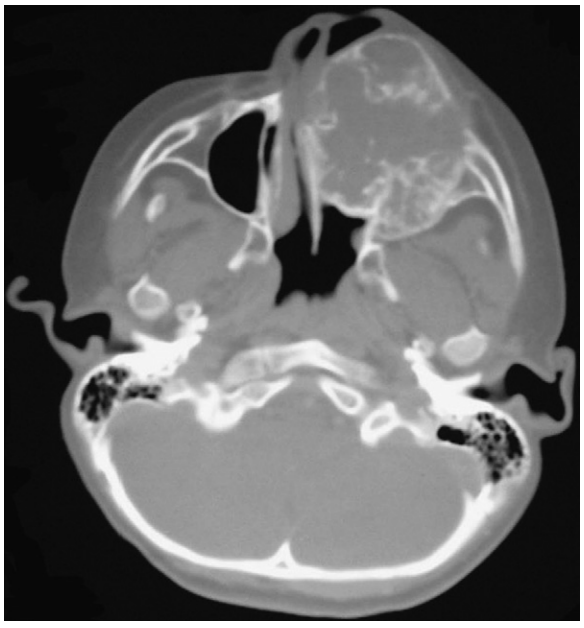

Fig. 1 Non-contrasted axial CT scan demonstrating a large maxillary sinus mass with facial distortion.

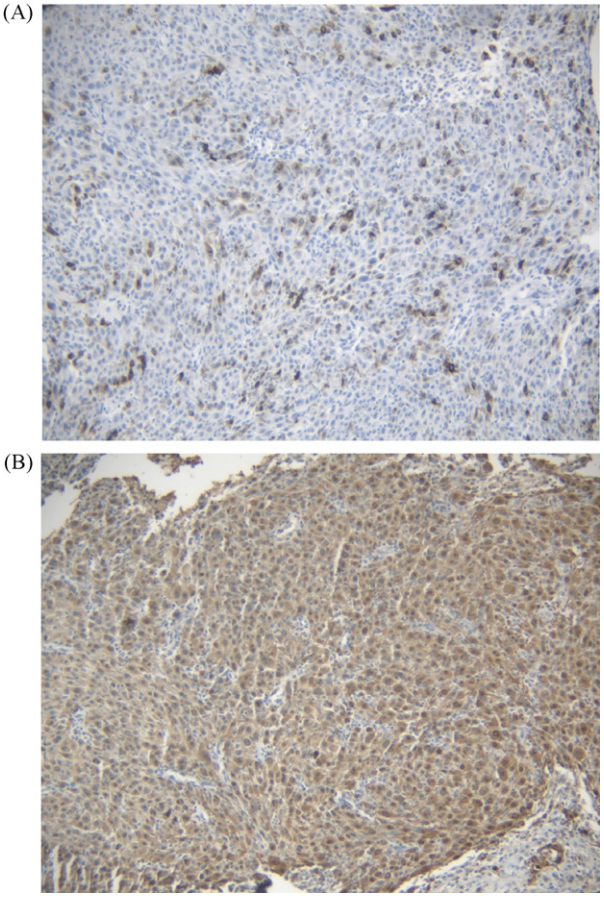

Fig. 2 Photomicrograph of the resected maxillary sinus melanoma. Immunohistochemical staining for HMB 45 (A) and S100 (B).

3. Discussion

In 1985 J. Aidan Carney described a group of patients who suffered from a complex of otherwise