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Case Report

When a stuffy nose won't go away: Rhabdomyosarcoma masquerading as adenoiditis ☆,☆☆

Daniel Griepp, BE[a,b], *Aron Soleiman, MD*[a], *Lisa Kurien, BS*[b], *Jessica Adukuzhiyil, BS*[b], *Abin Sajan, MD*[a,c,1,*]

[a] *Department of Radiology, SUNY Downstate Health Sciences University, 450 Clarkson Ave, Brooklyn, NY USA*
[b] *College of Medicine, New York Institute of Technology, 101 Northern Blvd., Old Westbury, NY, USA*
[c] *Department of Surgery, NYU Langone Health - Long Island, 259 1st Street, Mineola, NY, USA*

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ABSTRACT

Embryonal Rhabdomyosarcoma is a malignant mesenchymal proliferation of immature skeletal muscle and may arise in children in the orbit, middle ear, nasal cavity, paranasal sinuses, or nasopharynx. Clinical diagnosis may be difficult in a subset of patients who have no significant deformities or irregularities upon visual inspection of the oropharynx. Rhabdomyosarcoma in this setting may be mistaken for a more common underlying etiology such as an upper respiratory infection. We report a case of a 7-year-old male with embryonal variant rhabdomyosarcoma previously misdiagnosed by 3 different physicians to be adenoiditis based on clinical exam and laryngoscopy. This case highlights the capacity for rhabdomyosarcoma to mimic commonly encountered adenoiditis. It also serves as a reminder to maintain a high level of diagnostic vigilance and clinical suspicion of noninfectious etiologies when symptoms persist and are refractory to standard treatment.

Introduction

Rhabdomyosarcoma is a malignant proliferation of immature skeletal muscle [1]. It accounts for approximately 3% of all pediatric neoplasms and half of all solid malignancies in patients less than 10 years of age [2,3]. When rhabdomyosarcoma arises in the head or neck in children, nearly 50% are isolated to the middle ear, nasal cavity, paranasal sinuses, or nasopharynx [4,5]. Although rhabdomyosarcoma can present as generalized painless swelling, it may be accompanied with oculobulbar cranial nerve deficits, bloody

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* Corresponding author.
E-mail address: abin.sajan.01@gmail.com (A. Sajan).
[1] **Present address:** Department of Surgery, NYU-Winthrop University Hospital, 259 1st Street, Mineola, NY 11501, T: (718) 514-3022.
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discharge, dysphagia, and significant ear and nose pain due to mass effect on the nasopharynx and paranasal sinuses [2,4]. Computed tomography (CT) of the head and neck will show a mass and subsequent biopsy may confirm rhabdomyosarcoma diagnosis. However, this typically only occurs during a focused workup when a neoplasm is suspected. Diagnosis of rhabdomyosarcoma is challenging when symptomatology mimics a significantly more frequent underlying etiology, such as common seasonal allergies or recurrent upper respiratory infection [2]. Herein, we report a case of rhabdomyosarcoma that was previously misdiagnosed to be adenoiditis by 3 different physicians who relied on physical exam and laryngoscopy. A review of histological and immunohistochemical findings of rhabdomyosarcoma is also included along with a discussion of current treatment and long-term outcomes within the pediatric population.

Case description

An otherwise healthy 7-year-old boy presented to the emergency room complaining of intermittent fevers and noisy breathing. Three physicians had seen him independently over the course of 3 weeks at outside institutions for persistent, unexplained upper respiratory symptoms. Laryngoscopy had been performed and revealed significantly enlarged adenoids, but otherwise normal anatomy. He had been treated with antibiotics and steroids for presumed adenoiditis and was prescribed home Bilevel Positive Airway Pressure to improve breathing. However, his complaints of throat and ear discomfort with noisy breathing had progressively worsened. Physical exam revealed significant stridor and drooling while supine with resolution of these symptoms when the patient was placed upright. The patient's condition, in the setting of progressive symptomology refractory to antibiotic and steroid therapy, prompted a thorough workup.

CT imaging of the head and neck with contrast was performed and revealed a large 4.6 cm x 2.7 cm x 3.3 cm mass involving the right side of the nasopharynx. This extended laterally into the parapharyngeal space, superiorly to the skull base, and inferiorly to the level of the soft palate, shown in Figs. 1 and 2. The mass appeared to be obstructing the right eustachian tube with associated right mastoid disease. The right foramen ovale appeared larger than the left and bone destruction was noted along the inferior aspect of the right petrous apex. Mass effect on local structures may be seen in Fig. 2.

Given the patient history and imaging findings, the differential for the nasopharyngeal mass included nasopharyngeal carcinoma, dermoid cyst, and rhabdomyosarcoma. Surgical biopsy was performed, and subsequent pathological analysis revealed embryonal variant rhabdomyosarcoma. Immunohistochemistry was positive for desmin and actin. It was determined that the patient did not have concurrent adenoiditis, or adenoiditis secondary to rhabdomyosarcoma. Given the complicated location and large size of the tumor, surgical resection was delayed, and the patient was treated with combined chemotherapy of vincristine and dactinomycin with favorable recovery and resolution of symptoms at the 1-year follow-up.

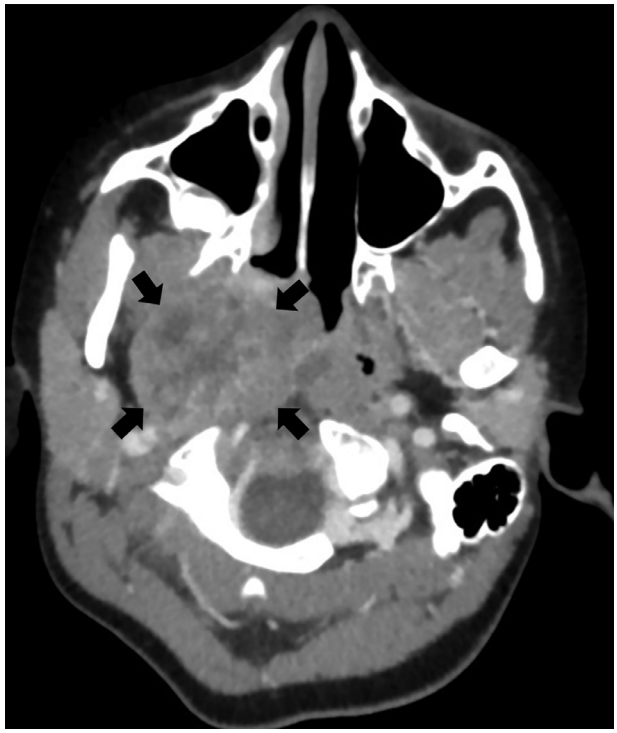

Fig. 1 – Axial view of contrast CT. Arrows show the well circumscribed, homogenous mass extending into the nasopharynx and laterally, into the parapharyngeal space. The mass appears to be obstructing the right eustachian tube with associated right mastoid disease. Bone involvement is also noted along the inferior aspect of the right petrous apex.

Discussion

Rhabdomyosarcoma is a malignant mesenchymal proliferation of immature skeletal muscle [1,6]. Rhabdomyosarcoma accounts for 3% of all pediatric neoplasms and half of all solid malignancies in patients less than 10 years of age, serving as the most common soft tissue sarcoma in the pediatric population [3]. The 4 major subtypes of rhabdomyosarcoma, distinguished histologically, are embryonal, alveolar, pleomorphic, and sclerosing spindle cell type [4]. The overwhelming majority of cases that occur in the head, neck, and genitourinary tract are of the embryonal subtype, as exhibited by our patient. Nearly 50% of cases in the head and neck region may be isolated to the middle ear, nasal cavity, paranasal sinuses, or nasopharynx; however, an estimated 25-31% of cases arise in the orbit alone [4]. The remaining 25% of cases occur in other locations such as the scalp, pharynx, oral cavity, parotid, thyroid, or parathyroid glands [5].

Histologically, embryonal rhabdomyosarcoma is characterized by small, round, hyperchromatic cells coupled with scattered heterogeneous tadpole-shaped rhabdomyoblasts consisting of eosinophilic cytoplasm, cytoplasmic cross-striations, and displaced nuclei [1,7]. When studied immunohistochemically, rhabdomyosarcoma is almost universally

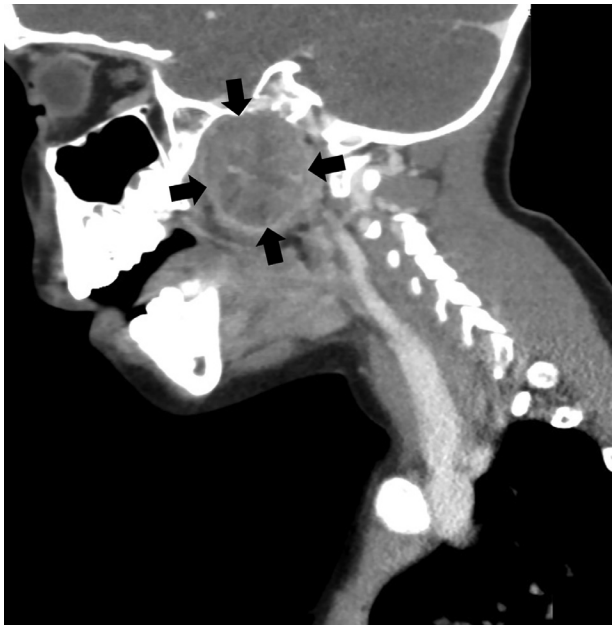

Fig. 2 – Sagittal view of contrast CT. Arrows show borders of well circumscribed, homogenous mass located in the superior nasopharynx, superiorly abutting the skull base, and posteroinferiorly adjacent at the level of the soft palate (likely attributed incorrectly to adenoiditis during laryngoscopy, leading to misdiagnosis).

positive for polyclonal desmin, whereas positivity for muscle-specific actin, myogenin, and myoglobin positivity is less reliable. Myogenin is well associated with the alveolar subtype and predicts a poor prognosis independent of tumor subtype [8,9]. Histologically, alveolar rhabdomyosarcoma appears as densely-packed ovoid tumor cells arranged between pseudo-alveolar spaces, vaguely resembling pulmonary alveoli. In order to diagnose alveolar rhabdomyosarcoma, the tumor must express greater than or equal to 50% of pseudo-alveolar architecture and a t(1;13) or t(2;13) translocation, known as a "FOXO1 rearrangement," identified by fluorescence in situ hybridization or reverse transcription polymerase chain reaction [6].

Staging of rhabdomyosarcoma requires identification of the primary site, size, nodal involvement, evidence of metastasis, and histological subtype. CT studies of the head and neck in the setting of rhabdomyosarcoma are useful to descriptively identify the location and tumor involvement in adjacent sites [10]. Common sites of metastasis, in order of frequency of involvement, include the lungs, bone marrow, bone, omentum, and pleura. Fewer than 25% of cases have overt signs of metastasis at the time of diagnosis [11].

The optimal approach for management of rhabdomyosarcoma in children includes a combination of chemotherapy, radiotherapy, and surgery [12,13]. Rhabdomyosarcoma is highly sensitive to chemo- and radiotherapy and current 5-year survival rates for embryonal and alveolar subtypes in pediatric patients have been found to range from approximately 65-91% [4,14,15] although some children may be blind due to orbital involvement [15]. If a tumor is found early, then a radical surgical approach is generally preferred due to preservation of function and cosmetic appearance. If found later at an advanced stage, chemotherapy is the best form of management. In the present case, given the complicated location of the tumor, surgical resection was delayed, and the patient was treated with combined chemotherapy of vincristine and dactinomycin.

The present patient had a confirmed embryonal rhabdomyosarcoma that was positive for desmin and actin. Genetic studies were not performed to assess for loss of heterogeneity or other abnormalities, but the patient's family history was not positive for rhabdomyosarcoma or any genetic syndromes. The tumor was in the nasopharynx, consistent with previously reported cases in the head and neck region. Although there was significant mass effect on the nasopharynx and the tumor was adjacent to the soft palate inferiorly and skull base superiorly, there was no indication of surrounding invasion or nodal involvement, confirming a lack of metastatic spread. This contributed to the favorable outcome with resolution of symptoms seen in the present case.

Although CT scans are generally avoided in pediatric patients, it is important to pursue further imaging in patients with refractory symptoms or those with an abnormal symptomatology. The risk of radiation exposure should be balanced with the diagnostic benefits of imaging.

Conclusions

This case highlights the capacity for rhabdomyosarcoma occurring in the head and neck region to mimic a commonly encountered upper respiratory tract infection. Thus, it is important to maintain a high level of diagnostic vigilance and clinical suspicion of noninfectious etiologies when symptoms persist over a prolonged period.

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