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Case Report

Beyond Adenoid Hypertrophy: Delayed Diagnosis of Nasopharyngeal Rhabdomyosarcoma in a Child

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Abstract

Persistent otorhinolaryngological (ENT) symptoms in children are usually attributed to benign conditions but may rarely conceal malignant diseases, leading to delayed diagnosis and poor outcomes. We report the case of a 3-year-old child who presented with progressive nasal obstruction and snoring, initially managed as allergic rhinitis and adenoid hypertrophy. Despite adenoidectomy with non-malignant histopathological findings, the child's condition deteriorated, with respiratory distress, weight loss, bilateral epistaxis, cervical lymphadenopathy, and neurological signs. Further investigations revealed an extensive nasopharyngeal mass with locoregional invasion and pulmonary metastases. Histopathological and immunohistochemical analyses confirmed embryonal rhabdomyosarcoma. Despite initiation of chemotherapy, the disease progressed rapidly, resulting in death. This case highlights the need for heightened clinical vigilance, early endoscopic evaluation, and multidisciplinary management when ENT symptoms persist or worsen in children.

Keywords: *Otorhinolaryngological (ENT); Adenoid Hypertrophy; Nasopharyngeal Rhabdomyosarcoma*

Introduction

Otorhinolaryngological (ENT) manifestations, such as snoring and nasal obstruction, are generally attributed to benign conditions. However, it is essential to recognize that a small proportion of these cases may be related to malignant etiologies.

In this article, we highlight the case of a young patient whose symptoms characteristic of nasopharyngeal rhabdomyosarcoma were initially misinterpreted as adenoid hypertrophy. This diagnostic error resulted in a significant delay in diagnosis and inappropriate management. Unfortunately, when the tumor diagnosis was eventually established, the disease had already progressed to a metastatic stage, ultimately leading to the patient's tragic outcome.

This case underscores the critical importance of clinical vigilance and the consideration of differential diagnoses when faced with similar symptom presentations. By sharing this observation, our aim is to raise awareness among healthcare professionals of the need to suspect a malignant etiology in the presence of persistent and atypical ENT symptoms.

Case Report (Observation)

This is the case of a 3-year-old patient with no significant past medical history, who presented with mild nasal obstruction eight months prior to admission. Over time, the obstruction progressively worsened without any medical consultation being sought. After four months, it became permanent, leading to constant and noisy mouth breathing, both day and night, which eventually prompted several medical consultations.

The patient was initially treated for allergic rhinitis and subsequently received multiple courses of corticosteroid therapy. Due to the lack of clinical improvement and the onset of snoring, a lateral nasopharyngeal radiograph was performed, revealing thickening of the nasopharyngeal soft tissues suggestive of adenoid hypertrophy with narrowing of the airway (Figure 1). An adenoidectomy was therefore performed. Histopathological examination of the adenoid tissue did not reveal any signs of malignancy. Consequently, all symptoms were attributed to adenoid hypertrophy.

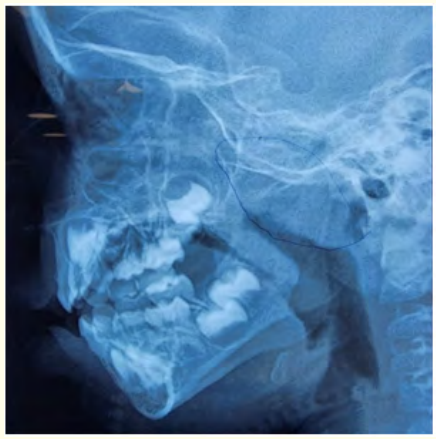


Figure 1: *Lateral nasopharyngeal radiograph of the patient.*

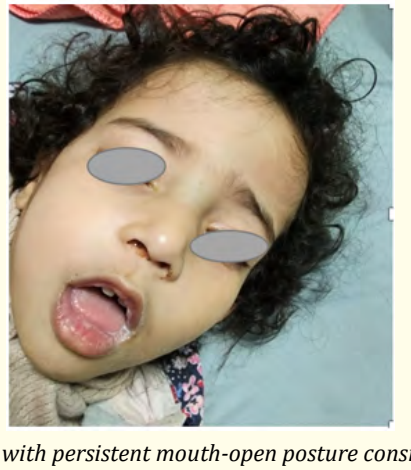
Despite this intervention, the patient's condition did not improve. She subsequently developed severe respiratory distress, significant weight loss, bilateral epistaxis, and purulent rhinorrhea. The patient eventually presented to the Specialty Hospital after a prolonged diagnostic delay, during which she had undergone eight consultations with a general practitioner, four with pediatricians, and three with otorhinolaryngologists, without a definitive diagnosis or clinical improvement (Table 1).

Timeline	Date	Healthcare professional	Decision / Management	Outcome
	01/06/23: Mild left nasal obstruction → no medical consultation → self-medication (family + pharmacists): antihistamines			No improvement
	01/10/23: Permanent bilateral nasal obstruction Mouth breathing Nocturnal snoring Preserved general condition			
1st medical consultation	03/10/23	General practitioner 1	Oral corticosteroid therapy + antihistamines (phenothiazine)	No improvement
2nd medical consultation	07/11/23	Pediatrician n1	Intranasal corticosteroids + antihistamines (cetirizine)	No improvement

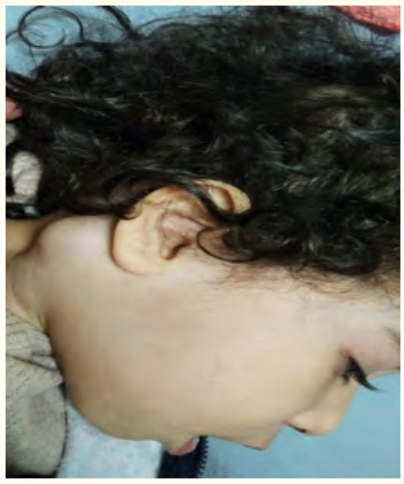
3rd medical consultation	27 /11 /23	Pediatrician n2	Intranasal corticosteroids + antibiotics (amoxicillin) → Referral to ENT specialist	No improvement
4th medical consultation	28/11/23	ENT specialist n1	Intranasal corticosteroids + antihistamines	No improvement + weight loss
5th medical consultation	12 /12/23	ENT specialist n1	Nasopharyngeal X-ray → Adenoid hypertrophy	Adenoidectomy scheduled after 10 days
6th medical consultation	22/12/23	ENT specialist n2	Blind curettage adenoidectomy Histopathology: no evidence of malignancy	No improvement + asthenia
7th medical consultation	29/12/23	ENT specialist n2	Clinical follow-up	Worsening snoring and noisy mouth breathing
8th medical consultation	08/01/24	ENT specialist n3	Clinical follow-up	
9th medical consultation	10/01/24	Pediatrician n3	Clinical follow-up	Clinical deterioration + 2 cervical lymph-adenopathies + bilateral epistaxis + purulent rhinorrhea + prolonged fever
10th medical consultation	30 /01/24	Pediatrician n4	Referral to the Specialty Hospital	
11th medical consultation	30/01/24	ENT specialist n4 – Specialty Hospital	Diagnosis of rhabdomyosarcoma	
	05/02/24	Pediatric Oncology (SHOP)	Admission to SHOP VAC chemotherapy cycle 1	
Death	10/03/24			

Table 1: *Timeline of medical consultations from the onset of symptoms to death.*

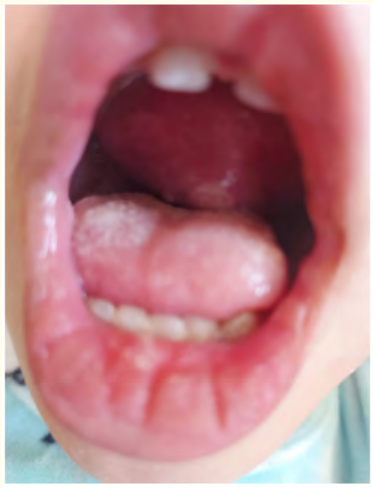
On clinical examination, she exhibited noisy mouth breathing and bilateral nasal obstruction with traces of epistaxis (Picture 1), bulging of both the soft and hard palate, and multiple cervical lymphadenopathies, including a right jugulocarotid lymph node measuring 4 cm in diameter, with a hard, fixed consistency and no signs of inflammation (Picture 2 and 3).



Picture 1: *Bilateral epistaxis with persistent mouth-open posture consistent with mouth breathing.*



Picture 2: *Clinical photograph showing right-sided cervical lymphadenopathy and persistent mouth breathing.*



Picture 3: *Large palatal mass.*

Neurological examination revealed right-sided ptosis and ipsilateral facial nerve palsy.

Nasofibroscopy revealed tumor tissue obstructing the nasopharynx and nasal cavities, with evidence of bone destruction. Histopathological examination confirmed the diagnosis of embryonal rhabdomyosarcoma (desmin positive, myogenin expression 10%).

Cervicofacial computed tomography demonstrated a mass measuring 44 × 77 × 55 mm, infiltrating the parapharyngeal spaces, maxillary sinuses, and both the hard and soft palate, with multiple bilateral cervical lymphadenopathies (Figure 2 and 3). Chest computed tomography revealed bilateral pulmonary nodules, consistent with metastatic involvement (Figure 4).

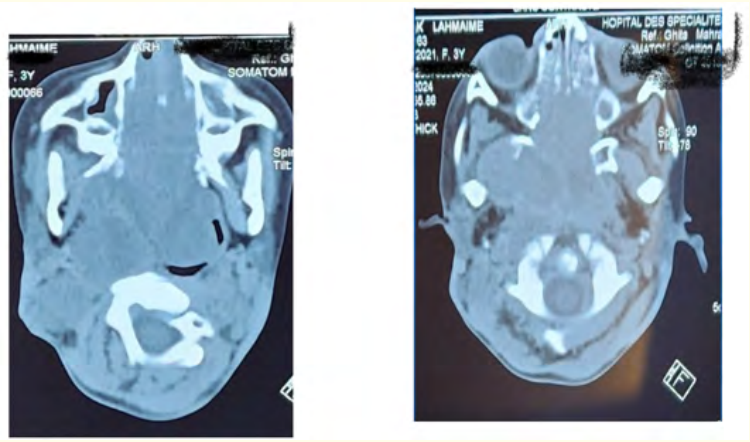


Figure 2 and 3: *Non-contrast cervicofacial CT scan showing an expansive nasopharyngeal mass with extensive locoregional invasion and multiple bilateral lateral cervical lymphadenopathies.*

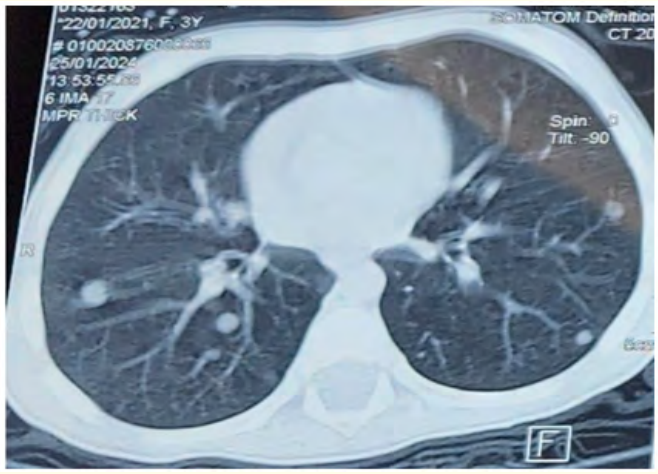


Figure 4: *Non-contrast chest CT scan showing multiple bilateral pulmonary nodules suggestive of pulmonary metastases.*

She was therefore immediately referred to the Department of Pediatric Hematology and Oncology.

In conclusion, this was a high-risk upper aerodigestive tract rhabdomyosarcoma, defined by the presence of several unfavorable prognostic factors: age under 10 years, tumor diameter greater than 5 cm, embryonal histological subtype, lymph node involvement, and the presence of metastatic disease. The clinical course was rapidly fatal, with the patient dying a few days after the initiation of chemotherapy, due to the extremely advanced stage of the disease at the time of diagnosis.

Discussion

This case highlights the critical importance of a thorough and rigorous evaluation of persistent ENT symptoms in children. Although nasal obstruction and snoring are frequently associated with benign conditions such as adenoid hypertrophy, persistence or worsening of symptoms despite appropriate treatment should systematically alert clinicians to the possibility of a more serious underlying etiology.

Our patient sought medical care on ten occasions from eight different physicians without a diagnosis of malignancy being established. She was repeatedly treated for allergic rhinitis and adenoid hypertrophy, which contributed to a major diagnostic delay and disease progression to a metastatic stage. This observation underscores several critical issues. First, the persistence of symptoms unresponsive to conventional treatment should prompt clinicians to consider more severe differential diagnoses. In this case, despite an initial interpretation favoring allergic rhinitis followed by adenoid hypertrophy, the progressive and persistent clinical signs should have triggered a more rigorous diagnostic reassessment.

Lateral nasopharyngeal radiography, often used as a first-line investigation, lacks both specificity and sensitivity. Recent studies confirm that it has no role in the diagnosis of adenoid hypertrophy [1,2]. In contrast, nasofibroscopy allows direct and detailed visualization of the nasopharynx, facilitating both the diagnosis of adenoids and the early detection of tumoral masses. In the present case, nasofibroscopy should have been considered as the initial examination, particularly in the presence of symptoms refractory to standard treatment.

Furthermore, a negative histopathological examination of adenoid tissue should not exclude a tumor diagnosis in the presence of a suggestive clinical presentation. Alarm signs such as respiratory distress, weight loss, epistaxis, and neurological involvement should prompt further in-depth investigations, regardless of initial findings.

Finally, a multidisciplinary approach involving ENT specialists, pediatricians, radiologists, and oncologists is essential for the rapid and appropriate management of complex and atypical presentations. This case illustrates the importance of specialist consultations and coordination among healthcare professionals to achieve an early and accurate diagnosis.

To better understand the importance of clinical vigilance, it is necessary to recall the main characteristics of rhabdomyosarcoma, a rare but aggressive malignant tumor that can affect the upper aerodigestive tract in children. It is the most common malignant soft tissue tumor in pediatrics, accounting for approximately 50% of such tumors and 5 - 8% of all childhood cancers [3]. Head and neck locations are predominant, representing up to 40% of cases [4,5].

Clinical manifestations include persistent nasal obstruction, epistaxis, cervical lymphadenopathy, visual or neurological disturbances, as well as general symptoms such as weight loss and asthenia. Diagnosis is primarily based on surgical biopsy with histological and immunohistochemical analysis. In rare cases, it may be established through cytological examination of pleural effusion or bone marrow aspiration.

Two main histological subtypes are described: the embryonal subtype, associated with loss of heterozygosity at chromosome 11p15.5, and the alveolar subtype, related to specific translocations t(2;13) PAX3-FOXO1 and t(1;13) PAX7-FOXO1 [6]. Microscopically, the embryonal subtype is characterized by high cellularity with undifferentiated mesenchymal cells, myxoid areas, and elongated or rounded "tadpole-shaped" cells. The alveolar subtype is characterized by small rhabdomyoblasts arranged in nests separated by fibrous septa, with a typical alveolar architecture [7].

Computed tomography and, especially, magnetic resonance imaging are essential to assess locoregional extension [8]. Treatment relies on a multimodal approach combining chemotherapy, radiotherapy, and surgery when feasible [9,10]. However, the effectiveness of this strategy largely depends on the timeliness of diagnosis.

In low-resource settings, early diagnosis remains a major challenge. Although specialized examinations such as nasofibroscopy, advanced imaging, and histopathological analysis are available, their implementation is often delayed due to late referral, diagnostic wandering, or initial underestimation of symptom severity. These delays contribute to silent disease progression toward advanced stages. It is therefore crucial to strengthen awareness among first-line practitioners regarding warning signs and to establish rapid referral pathways to specialized centers. Such strategies could significantly reduce diagnostic delays and improve the prognosis of children with nasopharyngeal rhabdomyosarcoma.

Conclusion

Nasopharyngeal rhabdomyosarcoma, although rare, should be considered in any child presenting with persistent or atypical nasal obstruction. This case emphasizes that diagnostic delay promotes progression to advanced stages and compromises prognosis. Increased clinical vigilance, early use of appropriate diagnostic tools, and rapid referral pathways are essential to improve survival outcomes in children affected by this malignant tumor.

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