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Nasopharyngeal Rhabdomyosarcoma Masquerading as Peritonsillar Abscess and Bronchopneumonia: An Unforeseen Diagnosis

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Abstract

Rhabdomyosarcoma is an aggressive malignant tumor and is the most common soft tissue sarcoma found in children, frequently arising from the nose, nasopharynx and paranasal sinuses. Here we report the case of a 3-year-old female who was misdiagnosed by multiple hospitals as peritonsillar abscess in view of the acute nature of the illness. Following tracheostomy due to the obstructive tendency of the lesion, the patient underwent imaging and biopsy which ultimately led to the diagnosis of a large nasopharyngeal rhabdomyosarcoma. The family was counselled regarding the prognosis and treatment modalities and the child subsequently underwent chemotherapy for the same.

Keywords

Rhabdomyosarcoma, Peritonsillar Abscess, Round Blue Cell Tumour, Pediatric Malignancy

1. Introduction

Rhabdomyosarcoma is the most frequently occurring soft tissue sarcoma in the pediatric population. Nearly 40% of these are localized in the head and neck region. Rhabdomyosarcoma constitutes a broader group of small round blue cell tumors (SRBCT)—a group of malignant low-grade tumors composed of small cells with large, round, hyperchromatic nucleus. They are a heterogeneous group of tumors, including neuroblastoma, rhabdomyosarcoma, Ewing's sarcoma, and non-Hodgkin lymphoma, desmoplastic small round cell tumor, medulloblastoma and retinoblastoma.

Rhabdomyosarcoma can be further classified into four subtypes—embryonal, alveolar, pleomorphic and spindle cell/sclerosing. Nearly 70% - 80% RMS constitute of embryonal subtype (ERMS) [1]. Most frequently diagnosed in the first 5 years, they can manifest at any age, even adulthood. ERMS was further classified into loose and dense types, of which the loose ERMS were subclassified into botryoid and non-botyroid, while dense ERMS into poorly differentiated and well-differentiated [2]. Alveolar rhabdomyosarcoma (ARMS) is more often found in adolescent and young adult population. These typically present as rapidly growing high stage tumors at extremities. Pleomorphic RMS (PRMS) is a high-grade sarcoma, presenting at a median age in the 6th to 7th decade [3]. PRMS needs to be morphologically differentiated from other high-grade tumors such as dedifferentiated liposarcoma and high grade undifferentiated pleomorphic sarcoma. Spindle cell or sclerosing rhabdomyosarcoma (SpRMS) is asymptomatic initially but may present with symptoms due to compression. These are well circumscribed lesions which show varying morphology.

It is not unusual for sinonasal RMS to present with symptoms mimicking allergic rhinosinusitis refractory to treatment. Persistence of symptoms calls for the need to conduct a detailed examination followed by appropriate radiological investigation, biopsy and histopathological analysis to confirm the diagnosis.

2. Case Report

A 3-year-old female child was brought to the department of ENT and head and neck surgery of our tertiary care hospital, referred from multiple hospitals for urgent intervention. On arrival, the child was on oxygen mask, had obvious stridor and intermittent episodes of apnea. Patient was brought with complaints of difficulty swallowing since 10 days and difficulty breathing since 2 days which were rapidly progressing. According to the history given by parents, the patient was admitted 2 months ago in a local hospital with complaints of difficulty breathing, fever, cough and coryza, which was diagnosed and managed as bronchopneumonia. However, the symptoms never completely resolved and general condition of the child deteriorated over the last 10 days. During this period, the patient was taken to multiple private hospitals where she was diagnosed with peritonsillar abscess and administered antibiotics. Initial WBC counts were mildly raised (14,000 cells/μL). Incision and drainage was planned in one of the hospitals, but the procedure was abandoned due to unknown reasons.

Patient had poor general condition, lethargic with an oxygen saturation in the range of 70 - 80 percent. Saturation further dropped in supine position.

On examination of oral cavity, soft palate appeared to be bulging downwards obstructing the entire oropharynx, with no visualization of either tonsils (**Figure 1**). Nasal cavity was filled with discharge. X-ray soft tissue nasopharynx was done which showed a large radio-opaque shadow obstructing the entire nasopharynx and oropharynx, approaching further into the neck (**Figure 2**). Patient was given IV steroids and an emergency tracheostomy was performed to achieve patent air-

way. Despite this patient was unable to maintain saturation and was shifted to the Pediatric ICU.

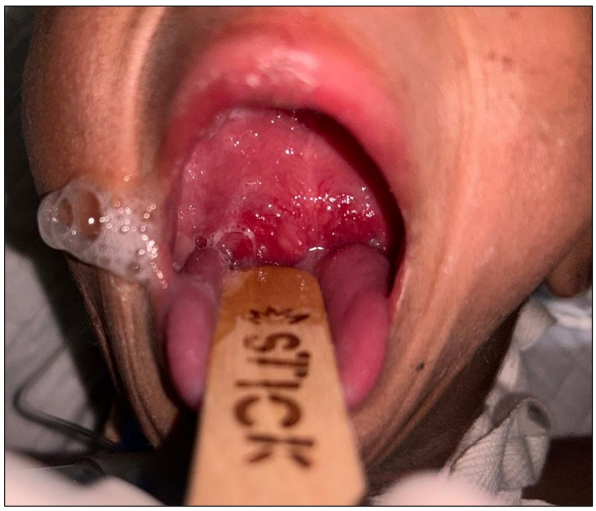

Figure 1. Oral cavity examination revealing bulging soft palate.

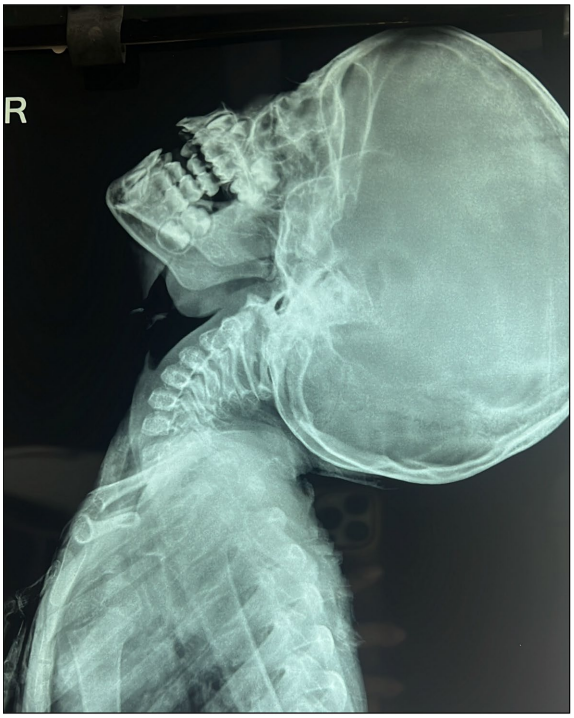

Figure 2. Radiograph showing lateral view of pharynx with radio-opacity in nasopharynx extending into oropharynx and hypopharynx.

Once stabilized, the patient underwent a contrast enhanced CT Head which showed a well defined iso-hypodense heterogenous enhancing lesion in left tonsillar fossa extending to cause complete blockage of oropharynx and extending superiorly into nasopharynx blocking bilateral nasal choana suggestive of tonsillar abscess (**Figure 3**).

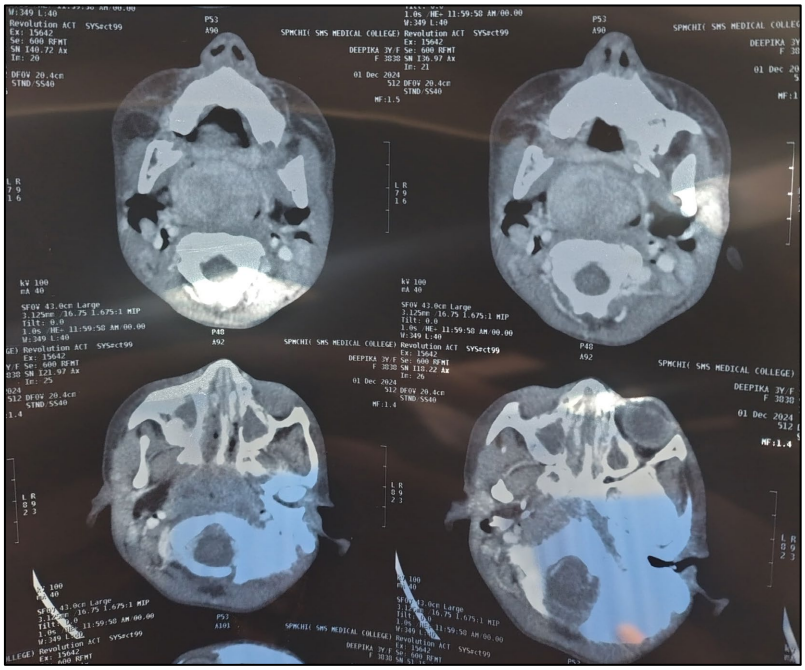

Figure 3. Computed tomography-axial section.

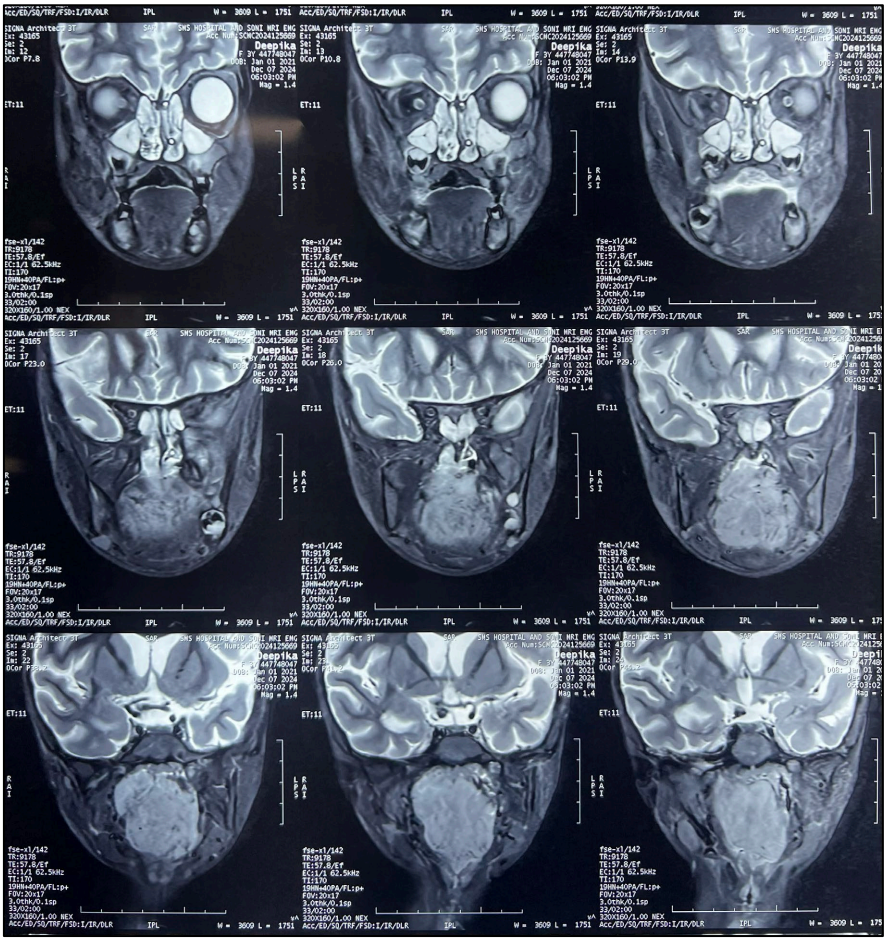

Figure 4. Magnetic resonance imaging (coronal section) showing mass lesion.

Wide bore needle aspiration was attempted, but no pus was aspirated. A small piece of tissue was collected and sent for biopsy. While the histopathology report was awaited, a contrast enhanced MRI was done in which the mass lesion was seen in nasopharynx and oropharynx, extending into hypopharynx, soft palate merging into the mass (**Figure 4**). The lesion showed moderate to intense enhancement on contrast suggestive of soft tissue mass with increased vascularity. Histopathology revealed fragments of tissue showing highly cellular lesion composed of closely packed sheets of small round dark hyperchromatic cells, scanty cytoplasm, vesicular nuclei with inconspicuous nucleolus and showing attempted rosetting as well as perithelial arrangement at places with frequent mitotic figures (**Figure 5**). Specimen gave the impression of a small round cell tumour. Following this immunohistochemistry was done which was immunopositive for Desmin and immunonegative for CD99 and Synaptophysin. Tumor cells were mildly positive for MyoD1 immunostain. A final diagnosis of rhabdomyosarcoma was made and the parents were counselled regarding chemotherapy. Patient received vincristine, actinomycin D and cyclophosphamide as chemotherapeutic agents. At 6 month follow up, complete remission was achieved, and patient was planned for decannulation of tracheostomy.

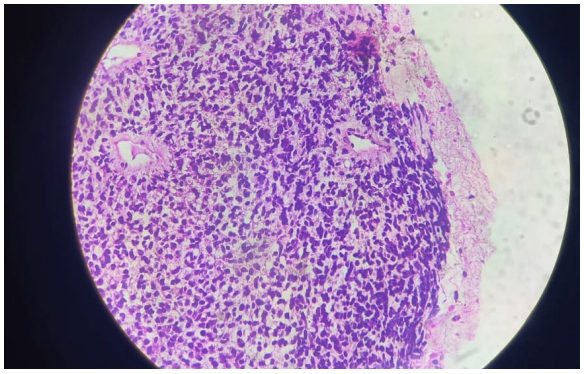

Figure 5. Histopathology suggestive of small round cell tumour.

3. Discussion

While the exact etiology of rhabdomyosarcoma remains largely unknown, evidence points to an underlying genetic component. Chromosomal translocations have been found in many tumors. RMS has been found to occur in many inherited cancer syndromes such as Noonan, Beckwith-Wiedemann, Costello and Li-Fraumeni syndrome [4] [5]. Other risk factors include fetal radiation exposure, drug use in parents, first-degree relative with history of RMS, pre-term birth, fertility drug use and history of congenital defects [6]. Symptoms of RMS in the head and neck region (nasopharyngeal, paranasal sinuses) can vaguely resemble allergic rhinosinusitis. As the disease progresses or involves orbit, symptoms such as proptosis, dizziness, nausea or headache begin to appear.

The histologic patterns and cytologic features of rhabdomyosarcomas range from undifferentiated small round blue cell neoplasms to lesions with advanced

cytohistologic features reminiscent of rhabdomyoma [7]. On histopathology, RMS can be differentiated from other small round blue cell tumors owing to positive staining for skeletal markers (myogenin, desmin and Myo-D1) and absence of differentiation. Shahidatul-Adha M. *et al.* reported the case of a 4-year-old girl who presented with acute onset squint secondary to sinonasal RMS and highlighted the rapidly progressive nature of the disease. In the review of literature by Shahidatul-Adha M. *et al.*, 9 confirmed cases of sinonasal RMS in pediatric population were reported, out of which only one case was of pleomorphic subtype, rest being embryonal RMS [8]. Almost all cases underwent chemotherapy. Whenever possible, tumor resection was performed via endoscopic approach, or debulking was done, followed by chemoradiotherapy, or solely chemotherapy.

Collaboration among pediatricians, oncologists, and radiologists is essential in determining the most effective treatment plan. Offering clear guidance and ongoing support to the parents is equally essential to avoid parents' refusal. A multidisciplinary and multimodal approach—including aggressive chemotherapy, radiotherapy, and surgical tumor debulking—has been shown to promote remission, enhance survival rates, and reduce both morbidity and mortality [9].

4. Conclusion

This case report highlights the importance of including nasopharyngeal RMS in the differentials of nasal masses. Understanding the broad clinical spectrum of this condition and the variation in treatment and prognoses with changes in morphological subtype is also brought to light. Its rare occurrence and aggressive nature emphasize the importance of continued and improved study regarding this tumor, so that agreed guidelines and a thorough understanding can be made which will guide rationale-based treatment.

Ethical Considerations

Written informed consent was taken from the parents of the patient for publication of case report along with clinical images.

Agreement to Conditions

All authors have read and approved the final version of the manuscript and agree to be accountable for all aspects of the work, ensuring its accuracy and integrity. The authors also confirm that the manuscript is original, has not been published previously, and is not under consideration or review by any other journal in the same or a substantially similar form.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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