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Case Report

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Pediatric Nasosinusal Rhabdomyosarcoma: Clinical Presentation and Therapeutic Challenges

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Abstract

Embryonal rhabdomyosarcoma (RMS) of the nasosinusal region is an uncommon pediatric malignancy, with an estimated incidence of 4.3 cases per million children annually. This report presents a case of embryonal nasosinusal RMS in a 12-year-old child managed at our institution, alongside a review of the literature. We discuss the epidemiology, diagnostic challenges, and therapeutic approaches associated with this rare tumor, providing insights to enhance clinical management and improve prognosis in pediatric patients.

1. Introduction

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children, originating from primitive mesenchymal cells with skeletal muscle differentiation. The nasosinusal region is an uncommon primary site, representing a diagnostic challenge due to nonspecific symptoms that mimic benign inflammatory conditions. Treatment strategies are multimodal, involving chemotherapy, surgery, and radiotherapy. Prognostic factors include tumor histology, size, location, and metastatic spread. This report describes a case of embryonal RMS of the nasosinusal region and reviews recent advancements in diagnosis and management.

2. Case Report

A 12-year-old male with no significant medical history presented with progressive bilateral nasal obstruction and purulent rhinorrhea persisting for five months despite medical treatment. Physical examination revealed an obstructive mass in the right nasal cavity without cervical lymphadenopathy or cranial nerve involvement. Imaging studies, including sinus CT, showed bilateral nasosinusal polyposis. MRI confirmed a Type IV nasosinusal lesion with posterior extension to the nasopharynx (figure 1,2). The patient underwent polypectomy, and histopathological examination confirmed embryonal RMS, botryoid subtype. Immunohistochemistry demonstrated positivity for desmin and myogenin, with negativity for caldesmon (figure 3). A metastatic workup, including thoraco-abdomino-pelvic CT and cerebrospinal fluid analysis, revealed no distant disease. The patient was classified as high-risk, subgroup E, and enrolled in the RMS 2005 protocol. He received three cycles of vincristine, actinomycin D, and ifosfamide (IVA) with good clinical and hematological tolerance. Treatment evaluation after three cycles guided the decision for locoregional therapy (radiotherapy ± additional surgery) and six additional cycles of IVA every three weeks. At treatment completion, the patient achieved complete remission, with no evidence of disease recurrence after ten years of follow-up.

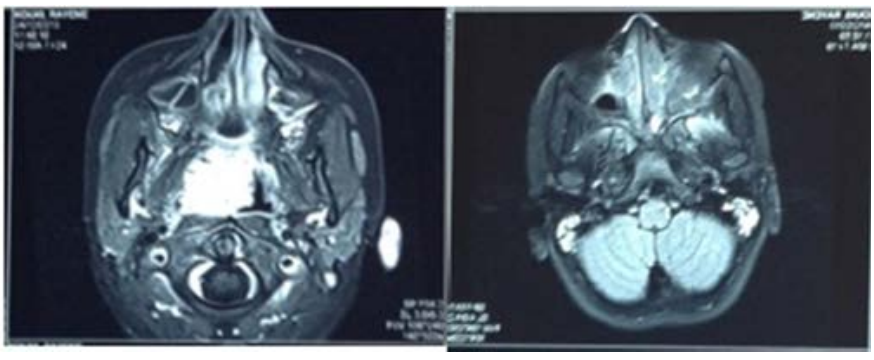

Figure 1,2: MRI of the Facial Mass Showing the Tumor Developing with Significant Posterior Extension Towards the Nasopharynx.

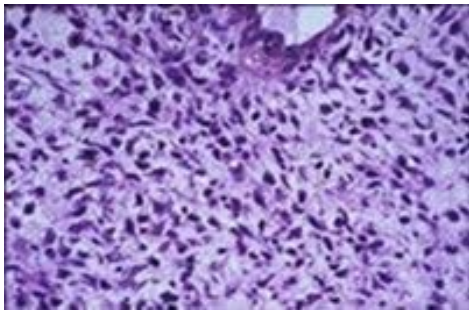

Figure 3: Microscopic Appearance of Embryonal Rhabdomyosarcoma (Rms)

3. Discussion

Rhabdomyosarcoma (RMS) remains one of the most common soft tissue sarcomas in children, accounting for approximately 60-70% of all pediatric cases. Nasosinus RMS, though rare, presents particular diagnostic and therapeutic challenges due to its deep location and often nonspecific clinical presentation [1,2]. This section discusses recent advances in the management and understanding of pediatric RMS, particularly focusing on its nasosinus form. Epidemiology and Risk Factors: Recent studies have shown that the annual incidence of RMS in children remains stable, with about 4.3 cases per million children per year, although incidence rates vary by region and age group. The peak incidence occurs in children under 10 years old, with a notable increase in cases between 1 and 4 years of age [3]. Recent data also suggest that alveolar RMS, associated with specific chromosomal translocations (t (2;13) (q35; q14) and t (1;13) (q36; q14)), is more commonly seen in adolescents, and is associated with a less favorable prognosis compared to the embryonal forms [4]. Genetic mutations continue to play a key role in understanding RMS risk factors. Syndromes such as neurofibromatosis type 1 and Li-Fraumeni syndrome remain well-established risk factors, but recent studies have also highlighted mutations in PAX3 and PAX7 genes, which are now considered important diagnostic and prognostic markers for nasosinus RMS [5,6]. Diagnosis and Staging: The diagnosis of nasosinus RMS in children relies on a combination of advanced imaging techniques, such as MRI and CT scans, along with histopathological confirmation.

Recent studies emphasize the superiority of MRI for detecting soft tissue tumors and evaluating regional extension, particularly in the nasopharynx and sinuses [7]. A 2023 study reported that MRI can more accurately detect peritoneal lesions compared to CT scans, making it a critical tool in the follow-up of these patients [8]. Immunohistochemical markers like desmin and myogenin are crucial for diagnosis, with high positivity rates in both embryonal and alveolar subtypes. Additionally, advances in cytogenetic testing have enabled the identification of specific translocations, such as the t (2;13), which is almost exclusively present in alveolar RMS and has been associated with a poorer prognosis [9]. Treatment and Prognosis: The treatment of RMS, including nasosinus RMS, has become increasingly targeted and multimodal. According to recent guidelines, treatment consists of a combination of chemotherapy, surgery, and radiotherapy, with an increasing role for targeted therapies and precision medicine [10].

Standard chemotherapy regimens, such as the IVA (Vincristine, Actinomycin D, Ifosfamide) protocol, remain widely used in high-risk forms, but new options, including anti-angiogenic therapies and mTOR inhibitors, are currently being explored in clinical trials [11,12]. A 2023 study reported that patients with botryoid embryonal RMS have 5-year survival rates exceeding 95%, which represents a marked improvement over more aggressive forms like alveolar RMS, which have 5-year survival rates between 50% and 60% [13]. The overall survival for patients with localized nasosinus RMS is generally better, but metastatic cases remain difficult to treat, with survival rates of 30-40% in these cases. Furthermore, newer radiation therapies, such as intensity- modulated radiation therapy (IMRT), are increasingly used to minimize side effects, particularly stereotactic techniques that improve precision in treating young children.

Prognostic Factors and Follow-Up: Prognostic factors for nasosinus RMS in children include age at diagnosis, tumor size, resecability, and the presence of metastases. Younger patients, particularly those under 10 years of age, tend to have a better prognosis, while older patients and those with larger, unresectable tumors present with poorer outcomes. A key factor identified in recent studies is chemotherapy resistance in alveolar RMS, leading to the exploration of new targeted therapies. Long-term follow-up remains essential, not only for detecting local recurrences or metastases but also for assessing treatment-related side effects, particularly on physical and neurological development (18). Follow-up protocols that include. Regular imaging with MRI and CT is recommended, as local recurrences remain a significant cause of mortality in patients treated for Naso sinus RMS.

Conclusion

Recent advancements in the understanding of Naso sinus RMS in children have significantly improved survival rates, especially for botryoid forms. Innovations in imaging, genetic diagnostics, and multimodal therapies continue to enhance the management of this rare tumor. However, further research is needed to optimize treatment for more aggressive forms and improve long-term outcomes for affected children.

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