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Case Report

Embryonal rhabdomyosarcoma presenting as edema of the soft palate and necrosis of the uvula: a case report and review of the literature

Oscar Calderon Wengerman[1], Jonathan Maya Epelstein[2]*, Hector De La Garza Hesles[3], Michelle Mizrahi Yaffe[4], María J. Sanabrais López[5]

[1]Department of Otolaryngology Head and Neck Surgery, Centro Médico ABC, Mexico City, Mexico
[2]Department of Pediatrics. Hospital Angeles Lomas, México
[3]Department of Otolaryngology, Head and Neck Surgery, Hospital Angeles Lomas, Mexico City, Mexico
[4]Universidad Panamericana, México City, Mexico
[5]Universidad Marista De Mérida, Mérida, Yucatán, Mexico

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***Correspondence:**
Dr. Jonathan Maya Epelstein,
E-mail: jmaya01@yahoo.com.mx

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ABSTRACT

Rhabdomyosarcoma (RMS) is a malignant tumor that affects skeletal muscles, typically found in the head and neck region of children. There have been a few reported cases of RMS occurring in the oral cavity. The prognosis of a patient with this condition depends on many clinical and biological factors, including age, the location of the tumor, histopathological subtype, and whether it has spread at the time of diagnosis. This case report and literature review focus on an unusual presentation of embryonal RMS in the uvula. The report described a case of a young Mexican child presented with symptoms such as edema, swelling of the soft palate, and necrosis of the uvula. The authors discuss the characteristics, management, and treatment of this neoplasm, providing valuable insights on the prognosis and therapeutic approaches for this rare condition and considering that the head and neck region can be a challenging site to treat due to the young age of the patient and the critical anatomy of this area.

Keywords: RMS, Embryonal RMS, Sarcoma, Soft palate, Uvula

INTRODUCTION

Rhabdomyosarcoma (RMS) is childhood's most common soft tissue sarcoma, accounting for approximately 3% of childhood malignancies.[1,2] These tumors are bimodal in distribution, often present in patients younger than 5 years and between 10 and 18 years.[3] Children aged 1 to 9 years have the best prognosis, while those younger than 1 year and older than 10 fare less well.[4] In addition to age, the primary tumor site plays an important role in the prognosis of the disease; therefore, it determines the therapeutic management strategy.[2,4]

RMS can appear in any body part, but it is more commonly found in certain locations such as the head and neck region (40%), genitourinary tract, and extremities.[2-4] RMS in the head and neck can be anatomically categorized into para-meningeal, orbit, and non-para-meningeal types.[5,6]

The first group comprises 25% of all RMS in the head and neck, including the nose, nasopharynx, paranasal sinuses, middle ear, mastoid, infratemporal fossa, and pterygopalatine fossa.[2] They have the worst prognosis because the tumors remain asymptomatic in the early stages and their resection is problematic.[2,4] The second category represents 9% of all RMS and has a 5-year survival rate of more than 90%, compared to the rate of 70% for patients with a para-meningeal tumor.[4] The prognosis of this type of tumor is better because symptoms are detected early. Less commonly involved

sites in the head and neck (non-para-meningeal) include salivary glands, larynx, middle and lower pharynx, thyroid gland, cheeks, and the oral cavity.[2-4] This last category is also linked to a positive prognosis.[4] However, due to the numerous critical structures surrounding the location of these tumors, treating this disease remains uniquely challenging.[5]

Clinically, symptoms may vary depending on the localization of the tumor. However, it usually presents as a rapidly growing mass, infiltrating neighboring tissues and spreading to lymph nodes and the distant organs.[2,7]

RMS is categorically defined as one of the "small round cell" tumors, with four distinct histological subtypes: embryonal, alveolar, spindle cell/sclerosing, and pleomorphic.[4,7] The embryonal type overwhelmingly prevails in children aged 0-4 and boasts the best prognosis, constituting 57 percentages of the all-RMS cases.[4]

RMS is often treatable in children with localized disease who receive combined therapy, with over 70% of patients surviving 5 years after diagnosis.[4] The intergroup RMS study (IRS) created a staging system based on stage, clinical group, and histology.[3,4] Low-risk RMS includes localized embryonal histology at a favorable site (such as the orbit) or localized disease of embryonal histology at an unfavorable site that has been grossly resected.[3,4]

The intermediate-risk group includes patients with localized embryonal histology at an unfavorable site with gross residual disease, as well as all patients with localized disease with alveolar histology. The high-risk group includes patients with metastatic disease.[3,4]

According to our review of the literature, only 5 cases have been reported in the indexed journals to date involving the uvula.[8-12] We report the first case in Mexico to our knowledge of an Embryonal rhabdomyosarcoma originating from the uvula.

CASE REPORT

A 1-year-old toddler was hospitalized with a 1-week history of snoring, hyaline rhinorrhea, nasal congestion, hypernasal voice, halitosis, and occasional bleeding from the mouth. Physical examination revealed a 2.5×3 cm red, firm mass with important edema and swelling of the soft palate with necrosis of the lower half of the uvula (Figure 1 A and B).

There were no signs of respiratory distress, however, due to the risk of airway occlusion, he was hospitalized for observation. Laboratory examinations, including the blood routine and C-reactive protein, revealed no abnormalities. Computed tomography confirmed the presence of an irregular mass partially obstructing the airway (Figure 2 and 3).

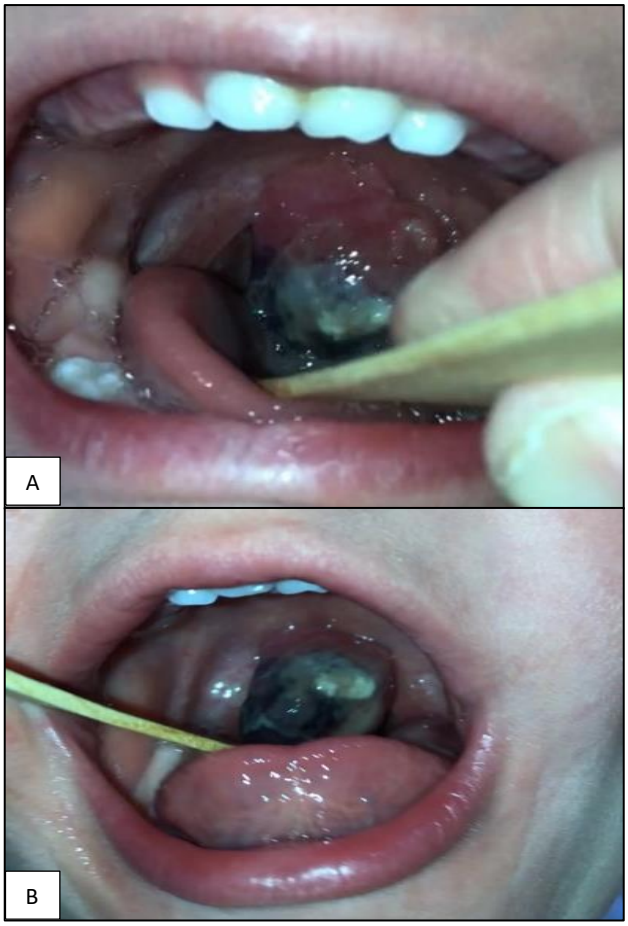

Figure 1 (A and B): Exophytic violaceus mass in the soft palate and uvula region. Necrotic inferior border of the mass.

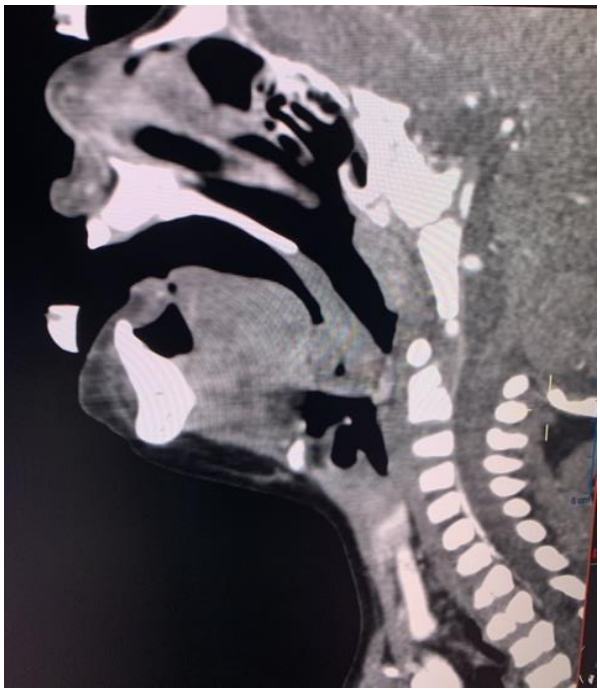

Figure 2: Sagital CT shows an irregular mass partially obstructing the airway.

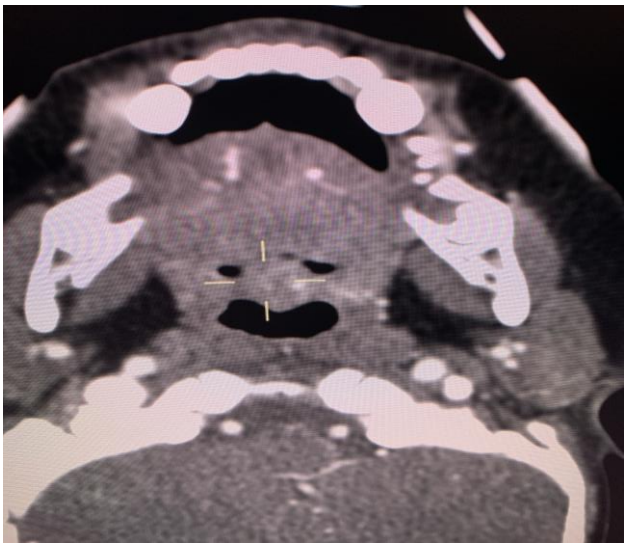

Figure 3: CT axial of tumor at level of uvula tumor.

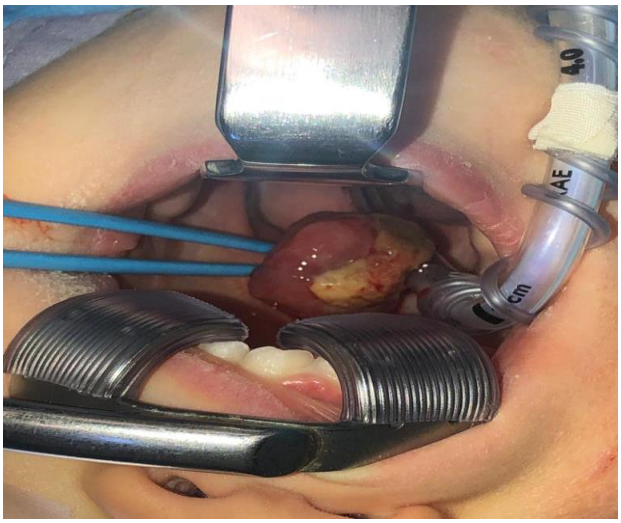

Figure 4: Intraoral resection of the tumor.

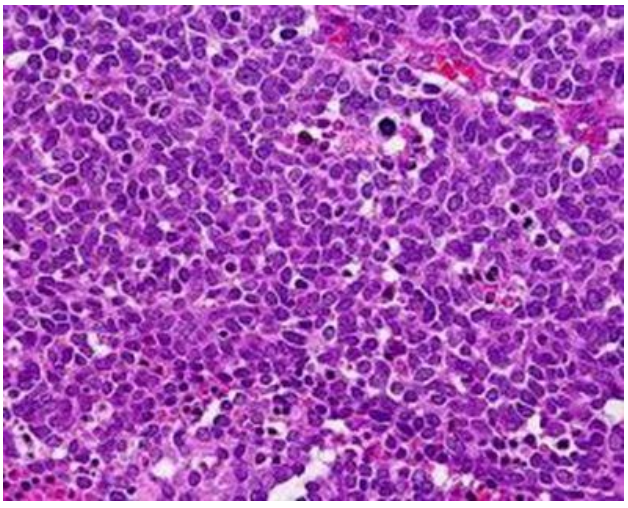

Figure 5: Embryonal RMS with a zone of stroma-rich, dense, small round cell appearance separated by small amounts of interstitial collagen.

Tumor was completely resected with no complications (Figure 4), and biopsy samples were obtained for definitive histopathological analysis, which reported small round blue cells with high-grade features (Figure 5). Neoplastic cells were strongly positive for CD 56+, vimentin, desmin, myoglobin, and muscle-specific actin. According to histopathological and immunochemical reports, it was diagnosed as a botryoid embryonal RMS with positive margins. Disease was categorized as group IIa according to IRS staging system; thus, tumor was classified as low-risk.

The patient started treatment at the memorial Sloan Kettering cancer center in New York city, which included 9 consecutive weekly doses of vincristine, 4 doses of dactinomycin and cyclophosphamide given at 3-week intervals (the VAC regimen), and a second 10-week block of chemotherapy consisting of 9 consecutive weeks of vincristine and 4 more doses of dactinomycin given at 3-week intervals with a 22-week protocol. Subsequently, local control was delivered using a novel radiation technique consisting of the administration of 30.2 Gy low-dose rate (LDR) brachytherapy with I-125. The patient completed treatment without severe adverse events and was discharged without complications.

DISCUSSION

RMS is a malignant neoplasm derived from primitive mesenchyme cells that undergo partial rhabdomyoblast differentiation and thus often arise at sites where skeletal muscle tissue is normally absent.[3] The incidence of RMS is the highest in children aged between 1-4 years and 10-14 years.[4] Males are 1.5 times more affected than females.[3,4] This case was unique because RMS was observed in a 1-year-old male.

Approximately 35-40% of childhood RMS occurs in the head and neck region, with involvement of the oral cavity being rare in 10-12% of the cases.[13] However, the cases reported in the literature involving the oral cavity indicate that most of them occur in the palate and tongue.[14] In our patient, the soft palate and the uvula were involved, this last is an uncommon location of RMS and therefore makes this case unusual.

In general, this tumor is usually fast-growing and infiltrating, and often appears as an enlarging, painless mass.[2,10,14] Patients with RMS may present signs and symptoms such as paresthesia, loss of teeth, trismus, and even severe respiratory obstruction.[13-15] Still, it depends on different factors such as advanced tumor stage, infiltrating growth, and tumor location.[4,13] In our case, when the patient presented to our clinic, he had vague symptoms like snoring, rhinorrhea, nasal congestion, halitosis, and occasional bleeding from the mouth, which appeared suddenly. Due to the patient's prompt consultation and the location and size of the tumor, we were able to remove it without complications, and the samples were obtained for definitive histopathological

analysis. However, surgical excision is challenging in cases of RMS of the oral region owing to the involvement of other crucial structures in this location.

Embryonal rhabdomyosarcoma is present in the majority of pediatric cases and includes botryoid and spindle cell subtypes.[4,13,14] Although the histopathological diagnosis of RMS can be difficult due to the similarity of this neoplasm to other small, round, blue-cell tumors of bone and soft tissue in childhood, in our case, the marked pleomorphism noted in the sample plus the immunochemistry-specific markers were sufficient to establish a precise diagnosis of Embryonal rhabdomyosarcoma.

Several immunohistochemical markers have been used for diagnosing RMS. Desmin and vimentin are considered prominent and useful markers to differentiate RMS from other soft tissue tumors, which tested positive in the tumor of the current case.[7]

Treatment of RMS continues to evolve, most patients undergo multidisciplinary treatment, as in the case of our patient, with surgery, chemotherapy, and radiotherapy.[4,14] In our case, RMS was diagnosed early, and appropriate treatment was initiated. There was no metastasis to other organs, the tumor was localized, and the histological type contributed to our patient's excellent prognosis.

CONCLUSION

We described the clinical, histological, and immunohistochemical aspects of a rare case of oral embryonal RMS. Although rare, soft tissue sarcomas should be included in the differential diagnosis of intra-oral lesions, especially in children so that they have prompt multidisciplinary care and a better long-term prognosis.

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