

## Rhabdomyosarcoma in the Oral Cavity of A Pediatric Patient: A Rare Case\*

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Dear Editor,

Rhabdomyosarcoma (RMS) is a rare, aggressive, and malignant neoplasm with rapid growth composed of primitive mesenchymal cells that exhibit skeletal muscle differentiation and mainly affects children and adolescents (60%) [1]. RMS is the most common soft tissue sarcoma with a rate of 50-60% in pediatric patients and ranks third among pediatric extracranial solid tumors, following Wilms tumor and neuroblastoma at a rate of 4-5% [2]. Head and neck localizations constitute 35-40% of cases, with oral lesions being extremely rare [3].

RMS has four well-defined subtypes: embryonal, alveolar, spindle cell/sclerosing, and pleomorphic. Embryonal RMS comprises 70-75% of all RMS cases [4], which are mostly sporadic. However, an increase has occurred in the number of recent studies on RMS, and the conclusions of these studies have shown that the children detected with defects in the RAS or Hedgehog pathways, as well as those with predisposing familial syndromes, carry a higher risk for developing embryonal RMS. In addition, PAX3-FOXO1 and PAX7-FOXO1 fusions have been identified in alveolar RMS and been accepted as diagnostic markers by many researchers [4-7].

The literature describes the clinical signs and symptoms of oral RMS as rapidly growing swelling, facial asymmetry, paresthesia, trismus, and difficulty swallowing [1,8].

Recently, we detected a rhabdomyosarcoma case in the oral cavity of a pediatric patient. The patient is a 6-year-old boy who was admitted to the pediatric clinic with a complaint of swelling

on the cheek. A mass lesion originating from the oral cavity was discovered during his clinical examination. Magnetic resonance imaging (MRI) showed a mass lesion that extended from the left maxillary sinus and posterior pterygopalatine fossa to the superior of the cavernous sinus and oral cavity. A part of the sphenoid sinus and left masseter muscle had also been infiltrated.

Three pieces of biopsy material were taken from the hard palate, with the size of the largest being 0.8x0.8x0.5 cm and the smallest being 0.7x0.5x0.2 cm. These were sent to our pathology laboratory due to the clinical pre-diagnoses of sarcoma, acinic cell carcinoma, and maxillary sinus tumor.

The histopathological examination showed a tumoral lesion formed by cells containing narrow eosinophilic cytoplasm, hyperchromatic round-oval nuclei arranged in the form of islands, and cords with a crush artifact in the fibrocollagenized stroma, including sporadic myxoid changes. Some cells with eccentric nuclei and some cells with spindle nuclei were also encountered infrequently. Atypical mitotic figures were frequently observed. No rosette formation was encountered.

Small round cell tumors, neuroendocrine tumor, plasma cell neoplasia, and malignant melanoma were included in the differential diagnosis. A large immunohistochemical panel including all differential diagnoses was performed. The immunohistochemical examination revealed the tumor cells to be positive for myogenin, desmin, and vimentin. The Ki67 proliferation index was ~80-90% per HPF. Tumor cells were negative for SMA, CD99, CD38, panCK, Synaptophysin, Chromogranin, CD3, CD20, S100, and MELAN-A.

\* This case has been submitted as a poster notification at 35<sup>th</sup> European Congress of Pathology, to be held from 9-13 September 2023 in Dublin, Ireland

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The patient was diagnosed with embryonal rhabdomyosarcoma. Because our hospital has no pediatric oncology unit, the patient was referred to another center for oncological evaluation and treatment.

The center to which the patient was referred classified the patient in the intermediate risk group according to the European pediatric soft tissue sarcoma study group (EpSSG). Embryonal RMS is treated via vincristine, actinomycin D, cyclophosphamide, and irinotecan.

RMS is among the small round cell tumors that occur in those of childhood age. Therefore, the differential diagnosis is quite extensive. In recent decades, nine cases of oral cavity RMS have been reported in pediatric patients [1, 9-12]. A pathological diagnostic evaluation is necessary for RMS because a specific diagnosis cannot be established based on clinical evidence alone [13]. Pediatric patients with rhabdomyosarcoma are classified in low-, intermediate-, and high-risk groups in accordance with EpSSG. Criteria have been defined such as the patient's age, tumor localization, histological subtype, presence of metastases at the time of diagnosis, with emphasis on prognostic importance [14].

Distinguishing between the subtypes has been recommended, because they may manifest differences in terms of biological behaviors and potential therapeutic options. By focusing on the histological features meticulously, however, reasonably selecting the differential diagnoses and correctly interpreting the immunostaining results can often allow us to achieve an accurate diagnosis, with a genetic analysis at times perhaps also being required.

In conclusion, this case has found worthy of presenting as a rare case in the literature.

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