DOI: 10.7860/JCDR/2025/78792.21137

Case Report

Ear, Nose and Throat Section

Multidisciplinary Approach in Management of Rhabdomyosarcoma of the Face: A Rare Case Report

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ABSTRACT

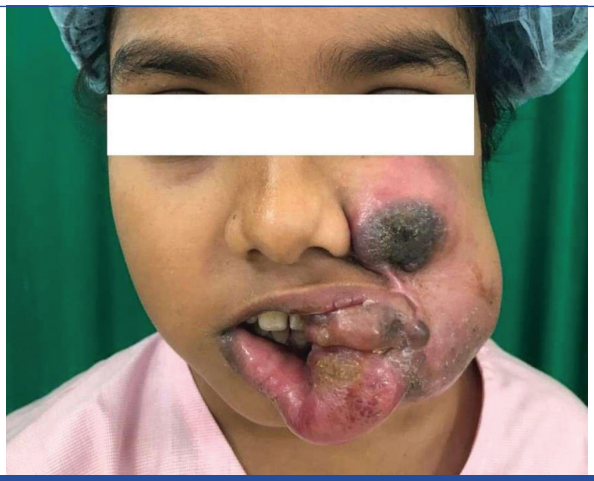
Rhabdomyosarcoma (RMS) is the most common soft-tissue sarcoma in children and adolescents. It arises from primitive mesenchymal cells that typically differentiate into skeletal muscle tissue. The causes and risk factors are not well understood, and most cases are sporadic, although some are linked to familial syndromes. A 10-year-old female patient presented with swelling on the left-side of her face. Following a wedge biopsy and histopathological and immunohistochemical analyses confirmed Embryonal Rhabdomyosarcoma (ERMS). Treatment involved a multimodal approach, combining surgical removal of the primary tumour, chemotherapy to address potential micro metastases, and radiotherapy for patients at higher risk. Improved survival rates are attributed to collaborative care and advancements in diagnosis and treatment. This case underscores the importance of coordinated care for timely diagnosis and effective treatment, leading to better patient outcomes and reduced morbidity.

Keywords: Chemotherapy, Embryonal type, Immunohistochemistry, Skeletal muscle tumour

CASE REPORT

A 10-year-old female patient was referred to a tertiary hospital with a complaint of swelling over the left-side of the face since one month. Similar lesions occurred on the left nasolabial region three months back, for which excision was done at a private hospital. The lesions were reported as dense subcutaneous fibromatous lesions, with a differential diagnosis of fibroma, neurofibroma, and keloid.

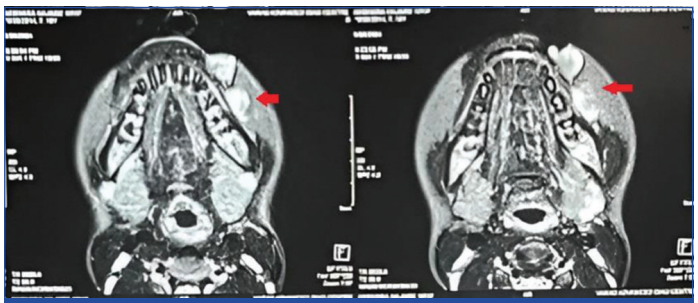
On examination, a solitary irregular swelling of size 7×4 cm was present on the left-side of the face involving the nasolabial region extending superiorly: 1 cm below the left lower eyelid, inferiorly: 1 cm above the mentum, laterally: at the level of left angle of mandible, medially: till the midline involving the left upper and lower lip and intraorally: till the lower second premolar and upper extent not visualised. There was a local rise of temperature, skin was erythematous, and hyperpigmented patches were seen [Table/Fig-1]. The swelling was firm to hard in consistency, tender and non-mobile, and no discharging fistula/sinus was seen. Left level 1b cervical lymph nodes and those on the left anterior aspect of the neck (probably 3 and 4) were palpable. In addition, two lymph nodes of size 2×2 cm present in the level 5 (5a and 5b) on the left were also palpable. All were approximately 2×2 cm in size, firm, mobile and non-tender. Provisional diagnosis was neoplastic lesion of the face.



[Table/Fig-1]: Pre-operative image.

Differential diagnosis was fibroma, neurofibroma and malignant lesions likely RMS. Ultrasonography of local swelling findings showed a diffuse, partly defined heterogenous hypoechoic lesion noted in the pre maxillary region on the left-side measuring approximately. 4.2×3 cm and 3×1.6 cm at an angle of the mandible on the left side, showing prominent vascularity. No calcification was noted within. It extended along the angle of the mouth inferiorly upto sub mandibular region and laterally upto pre-preauricular region displacing the parotid gland laterally. Multiple enlarged conglomerated lymph nodes with altered fatty hilum were noted bilaterally in the cervical region (1B,2,3), 5 on the left-side, the largest measuring 1.8×1.7 cm at level 2 and 1.9×1.7 cm at level 2B on the left-side.

Magnetic Resonance Imaging (MRI) (plain+contrast) of the face revealed a fairly well-defined two to three T1 hypointense and T2 heterogeneously hypointense lesions noted in the anterior maxillary region, near the upper part of the angle of mouth on the left-side (size approximately 2.0×1.5 cm) in the subcutaneous plane and another lesion noted in the lower part of the angle of mouth and extending posteriorly on the left side of the buccal surface (size approximately 3.4×1.1 cm). These lesions were seen involving adjacent fascia and muscles. On the post-contrast study, these lesions showed moderate heterogeneous enhancement. Multiple enlarged lymph nodes were noted in the bilateral submandibular regions and left level 2, 3, and 5 [Table/Fig-2].

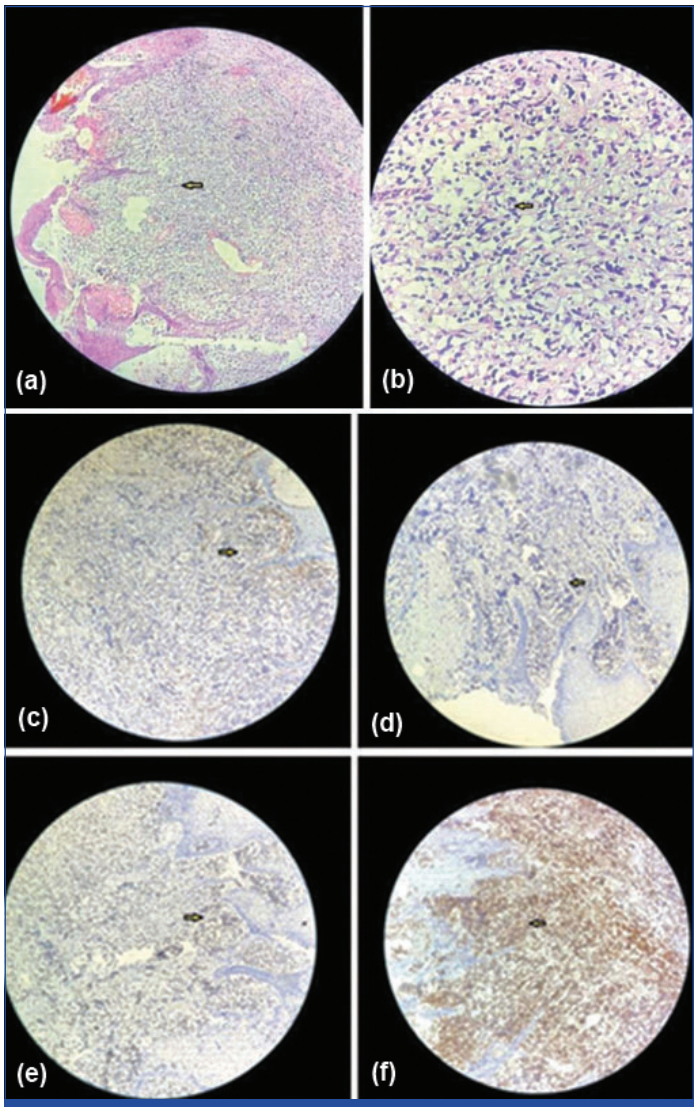


[Table/Fig-2]: MRI- arrows show fairly well-defined two to three T1 hypointense and T2 heterogeneously hypointense lesions in the anterior maxillary region, near the upper part of the angle of the mouth on the left-side.

Consent from relative (mother) was taken. The patient underwent an examination under anaesthesia followed by biopsy from the left facial and intraoral mass. General anaesthesia was administered

using fibreoptic flexible bronchoscopy as it was difficult to intubate. Fine needle aspiration cytology of the left submandibular lymph node was suggestive of Non-Hodgkin's lymphoma, morphologically suggesting Burkitt's lymphoma.

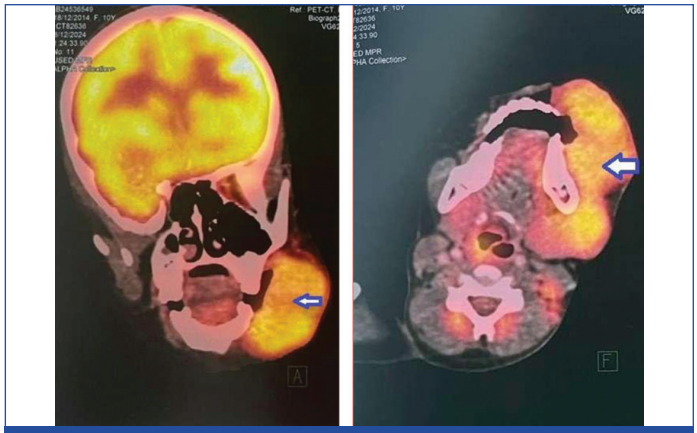
Histopathologic findings (biopsy) showed RMS, embryonal type. In immunohistochemistry, tumour cells were positive for vimentin, desmin, myo D1 and CD10. ATRx was retained in tumour cells. Ki67 labelling index was approximately 50-60% and the lesion was confirmed as RMS, embryonal type [Table/Fig-3].



[Table/Fig-3]: (a) The arrow indicates subepithelium showing tumour (haematoxylin and eosin, 100X); (b) The arrow shows spindle shaped cells (haematoxylin and eosin, 400X); (c-f) Showing tumour cell positive for vimentin, desmin, myoD1 and CD10 (Arrows represent the cells that have been stained by vimentin, desmin, myoD1 and CD10, respectively).

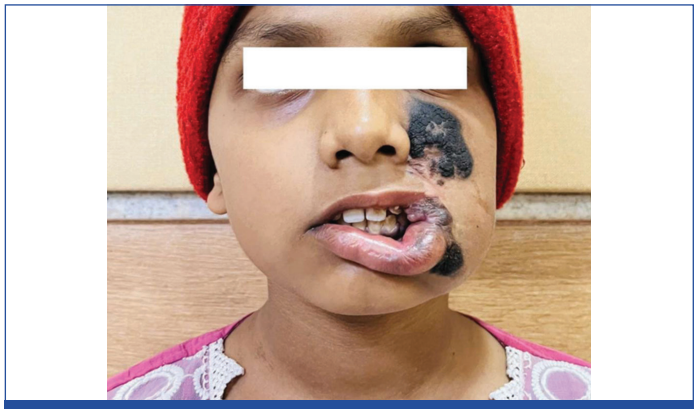
When consulted with a medical oncologist and radiation oncologist, positron emission tomography scan was performed, which was suggestive of a hypermetabolic large soft-tissue mass involving the left buccinator-buccal mucosa complex, representing the site of primary malignancy. Another smaller hypermetabolic mass in the left pre-maxillary region was part of the same disease. Several small and enlarged hypermetabolic left cervical level 1B-4 lymph nodes appeared metastatic [Table/Fig-4]. Two hypermetabolic discrete nodular lesions in the right lung lower lobe were likely to be metastatic. Few other irregular patchy fibro- consolidatory opacities in the bilateral lungs with minimal to low grade metabolic activity were possibly infective changes. Few hypermetabolic enlarged mediastinal lymph nodes indicated that the possibility of metastasis could not be completely ruled out.

Patient was sent for chemoradiation therapy at a tertiary cancer centre after seeking opinion from the paediatric oncologist. She



[Table/Fig-4]: PET-scan images- The arrows show hypermetabolic large soft-tissue mass involving the left buccinator-buccal mucosa complex that represent the site of primary malignancy. Another smaller hypermetabolic mass in the left pre-maxillary region is a part of the same disease.

completed three cycles of chemotherapy (Vincrystine, Endoxan, Actinomycin-D, Mesna and Peg G-CSF). Patient showed signs of improvement and swelling size reduced gradually [Table/Fig-5].



[Table/Fig-5]: Image showing decrease in size after three cycles of chemotherapy.

DISCUSSION

RMS was first noted in the mid-19th century, with early descriptions of similar conditions [1]. RMS is a highly aggressive tumour known for its rapid growth, primarily affecting children. It tends to occur in two main age groups: between 1-4 years and 10-14 years [2]. There is gender disparity, with males being approximately 1.5 times more likely to develop RMS than females [2].

The case report by Shrutha SP and Vinit GB describes a one-year-old boy with oral alveolar RMS presenting as a rapidly enlarging, painful swelling in the upper left maxillary ridge, which progressed to significant bone destruction and facial asymmetry within months. Despite a multidisciplinary diagnostic approach- including clinical, radiological, histopathological, and immunohistochemical assessment- the outcome was poor due to incomplete treatment; the parents refused radiotherapy, and the child developed bone and lung metastases, resulting in death six months later. This case underscores that paediatric RMS can present as an aggressive oral mass with extensive local invasion and a high-risk of metastasis, especially in very young children. Early, accurate diagnosis and a full course of multimodal therapy are critical for improving prognosis, as incomplete or delayed treatment significantly worsens outcomes [3].

Prognostic factors such as age, histologic subtype, and chemotherapy response are closely linked to survival [4]. Children older than three years and those achieving complete remission have far better outcomes, while younger children and those with recurrence or incomplete response have much worse outcomes. Five-year overall survival rates for paediatric head and neck RMS can reach up to 91% with comprehensive treatment, but these rates drop sharply in cases of recurrence, metastasis, or treatment resistance [4].

This case underscores the aggressive nature of recurrent paediatric head and neck RMS and the urgent need for novel therapies and improved risk stratification, as even aggressive, comprehensive management may not prevent rapid progression in high-risk or refractory tumours [4].

ERMS is characterised by a histological appearance that mirrors the embryonic stages of skeletal muscle development. The tumour cells are poorly differentiated, featuring small, round or oval shapes with hyperchromatic nuclei and minimal cytoplasmic definition [5]. In the present case, this lack of distinct histological features complicates the diagnosis, making additional immunohistochemical testing essential for confirmation.

The diagnosis of RMS is challenging due to its diverse histological presentations. It can manifest as a range of cell types, from small round cells to elongated, strap-like cells and large pleomorphic cells, often with sparse cross-striations. The presence of benign inflammatory cells can sometimes mask the neoplastic cells, further complicating the diagnostic process. Nonetheless, a poorly differentiated tumour without cross-striations, coupled with immunohistochemical analysis, can provide a definitive diagnosis of RMS [6].

To accurately diagnose RMS, careful histological and immunohistochemical assessments are necessary to rule out differential diagnoses such as Ewing's sarcoma, spindle cell sarcoma, and osteosarcoma [7]. Key immunohistochemical markers, including desmin and vimentin, are instrumental in identifying RMS and distinguishing it from other soft-tissue tumours [8]. In this case, the tumour cells were positive for desmin, vimentin, myoD1, and CD10.

The treatment of RMS involves a multidisciplinary approach, incorporating surgery, radiotherapy, and various chemotherapy regimens. Treatment choices are guided by the tumour's stage and clinical presentation. Important prognostic factors include the patient's age, tumour location, and histological subtype. For example, orbital tumours in the head and neck region are often managed with radiation therapy alone or in combination with chemotherapy. Non-orbital, non-parameningeal head and neck tumours are typically treated with surgery, which generally results in low long-term morbidity [9].

The initial management of RMS typically involves complete surgical excision of the tumour, aiming to preserve function and appearance. Radiation therapy and chemotherapy are used to control the tumour locally, reduce its size, and address tumours that are difficult to remove surgically [10]. However, these treatments can have significant side effects [11]. In the case presented, the patient received neoadjuvant chemotherapy with Vincristine, Cyclophosphamide (Endoxan), Actinomycin-D, Mesna and Peg G-CSF [12].

CONCLUSION(S)

Rapidly, progressing facial swelling in a paediatric patient should be considered as mostly sarcomatous malignancy. It should be meticulously evaluated by conventional histopathology and immunohistochemistry after proper clinical and imaging evaluation. Prompt and early action was taken by the ENT surgeon, anaesthesiologist, pathologist, and oncologist to come to a final diagnosis. The combined modality treatment given has benefited the patient. Periodic follow-up is necessary to see the response of treatment and chances of complications following chemoradiation in children.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]

- Plagiarism X-checker: Mar 01, 2025
- Manual Googling: May 01, 2025
- iThenticate Software: May 03, 2025 (5%)

ETYMOLOGY: Author Origin

EMENDATIONS: 6

AUTHOR DECLARATION:

- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Feb 27, 2025**
Date of Peer Review: **Apr 19, 2025**
Date of Acceptance: **May 05, 2025**
Date of Publishing: **Jun 01, 2025**