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RESEARCH ARTICLE

RHABDOMYOSARCOMA–THE ENIGMATIC SOFT TISSUE SARCOMA OF CHILDHOOD: A CASE REPORT WITH DIAGNOSTIC INSIGHTS

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Abstract

Embryonal Rhabdomyosarcoma(ERMS) is a rare,aggressive malignanc yoriginating from skeletal muscle progenitors.This case report describes a unique presentation of ERMS in an 8-year-old girlwho reported with a rapidly progressing swelling on the right side of her face of one-week duration, initially suspected to be due to trauma. An intraoral swelling on the right side of hard palate region with pathological mobility of the molar teeth was noted. A comprehensive diagnostic workup,including imaging and biopsy, was done.Histopathol ogical evaluation revealed the characteristic features of ERMS of the hard palate.Early diagnosis is critical in reducing the mortality rate of rhabdomyosarcoma. Individuals with delayed diagnosis were found to have a higher incidence of advanced-stage cancers and increasingly poor outcomes. This report discusses the importance of early recognition and accurate diagnosis in improving clinical outcomes.

Introduction:-

Sarcomas comprise of a heterogeneous group of malignant tumors that arise in the soft tissues or bone. Soft tissue sarcomas (STS) are derived from primitive mesenchymal cells such as muscle, connective tissue (tendons and synovial tissue), supportive tissue (fat and nerves), and vascular tissue (lymph and blood vessels). Rhabdomyosarcoma (RMS) is the most common STS in younger children, comprising more than half of these tumours in children up to 9 years of age.[1] Rhabdomyosarcoma accounts for approximately 7.5% of all childhood neoplasms. It is a relatively rare malignancy, with an annual incidence of about 8 cases per 1 million children under 16 years of age. The disease predominantly affects younger age groups, with nearly 70% of cases diagnosed in children below 10 years of age. A slight male predominance has been observed, with a male-to-female ratio of 1.4:1.[2]. Most cases of rhabdomyosarcoma occur sporadically, with no identifiable inherited cause. However, a subset of patients exhibits a genetic predisposition for the disease. One such example is Li-Fraumeni syndrome, which is associated with mutations in the p53 tumour suppressor gene (TP53). Families with this mutation often show a higher incidence of brain tumours, breast carcinomas, and adrenocortical carcinomas, alongside an increased likelihood of rhabdomyosarcoma in children.[3]

Environmental and prenatal factors may also contribute to risk. Children with Fetal Alcohol Syndrome and those born to mothers who used marijuana or cocaine during pregnancy have been observed to have a higher susceptibility to rhabdomyosarcoma.[2]

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The most commonly affected areas are the head and neck region, genitourinary tract, retroperitoneum, and, to a lesser extent, the extremities. The head and neck RMSs are anatomically divided into 2 categories: Para meningeal (including RMS of the nose, nasopharynx, paranasal sinuses, middle ear, mastoid, infratemporal fossa, and pterygopalatine fossa) and non-parameningeal (including RMS of the scalp, orbit, parotid gland, oral cavity, oropharynx, and larynx). RMS of the oral cavity accounts for 10–12% of all the head and neck RMS cases.[4] The current World Health Organization classification divides RMS into the following 4 subtypes: embryonal (ERMS), alveolar (ARMS), spindle cell/sclerosing (SSRMS), and pleomorphic, and combines morphology and molecular genetics.[5]This case report describes a unique presentation of ERMS in an 8-year-old patient, highlighting the diagnostic challenges and strategies employed to reach a definitive diagnosis.

Case Report:-

History and examination:-

An 8-year-old girl with no significant past medical or family history presented with a rapidly progressive swelling on the right side of the face of one week's duration. The child had a history of facial trauma sustained while playing, approximately one month before presentation.

Extraoral examination revealed a diffuse, firm, and tender swelling involving the right cheek. Intraorally, a pulsatile palatal swelling measuring approximately 50 mm × 40 mm was observed in relation to the maxillary right primary molars and the permanent first molar. On palpation, there was obliteration of the buccal vestibule along with expansion of both buccal and palatal cortical plates (Figure 1).

The right maxillary primary molars and permanent first molar exhibited marked mobility. The oral hygiene of the patient was good, and no dental caries were noted on any teeth. There was no regional lymphadenopathy.

Imaging:-

An orthopantomogram (OPG) obtained during the initial presentation revealed a diffuse radiolucent lesion involving the right maxilla and maxillary sinus, with displacement of the developing premolar tooth buds (figure 2).

To further assess the extent, borders, and nature of the lesion, a cone beam computed tomography (CBCT) scan was performed.

The CBCT demonstrated an extensive expansile lesion of the right maxilla, measuring approximately 46.85 × 49.96 × 35.19 mm in dimension (figure 4).Superiorly, the lesion extended upwards, displacing the floor of the right orbit and causing thinning of the orbital bone (figure 5).Superomedially, there was a disruption in the continuity of the junction between the medial wall and floor of the right orbit (figure 5).Medially, the lesion had destroyed the medial wall of the maxillary sinus and extended into the right nasal cavity, resulting in leftward deviation of the nasal septum.Inferiorly, there was alveolar bone destruction in relation to teeth 14, 15, and 16, producing a characteristic “teeth floating in air” appearance.Buccopalatally, the lesion exhibited bicortical expansion with destruction of the palatal bone up to the midline.Posteroinferiorly, it extended to involve the right pterygoid plates, and posteriorly, had destroyed the posterior wall of both the maxillary sinus and maxilla.Laterally, the lesion had eroded the posterolateral wall of the right maxillary sinus.

The radiological features were suggestive of an aggressive benign neoplasm, with differential diagnosis that included malignant tumor of the right maxilla or a high-flow vascular lesion (figure 3.a and 3.b).

Histological diagnosis:-

Fine-needle aspiration cytology (FNAC) from the right posterior maxillary region revealed the presence of red blood cells with scanty inflammatory cells, but was inconclusive for definitive diagnosis. Consequently, an incisional (Tru-cut) biopsy was obtained through a buccal vestibular approach under general anesthesia to establish the histopathological nature of the lesion.

Hematoxylin and eosin-stained sections demonstrated spindle-shaped tumor cells arranged in fascicles. The tumor cells exhibited ovoid to elongated “cigar-shaped” hyperchromatic nuclei with scanty cytoplasm. Frequent mitotic figures were also noted, indicating a high proliferative activity.

On immunohistochemical analysis, the tumor cells showed strong cytoplasmic positivity for Desmin, confirming skeletal muscle differentiation. Additionally, nuclear positivity for MyoD1 was observed, further substantiating the skeletal muscle lineage of the neoplasm.

Based on the histopathological and immunohistochemical findings, the lesion was diagnosed as a malignant spindle cell neoplasm, most consistent with rhabdomyosarcoma of the hard palate. Considering the anatomical site and histological pattern, the final diagnosis was established as a parameningeal embryonal rhabdomyosarcoma.

Following the confirmation of diagnosis, the patient was referred to the Regional Cancer Centre for specialized oncologic management. A combined chemotherapy protocol was initiated, consisting of Vincristine, Actinomycin D, Cyclophosphamide, and Dexamethasone, followed by radiotherapy.
Despite the initiation of multimodal therapy, the disease demonstrated an aggressive clinical course, and the child unfortunately succumbed to the illness approximately 2.5 months after the initial presentation.

Discussion:-

The incidence of RMS is the highest in children aged 1–4 years, lower in children aged 10–14 years, and lowest in those aged 15–19 years. These tumors exhibit a fast and aggressive growth, reaching large dimensions, and are generally painless, associated with high rates of recurrence and generalized metastases through the hematogenic and/ or lymphatic routes.[6]Oral rhabdomyosarcomas usually present as a rapidly enlarging painless mass and are generally larger than 1 cm at initial presentation. Involvement of adjacent nerves might cause paresthesia and pain. The tongue is the most common site involved in oral rhabdomyosarcomas, followed by the palate and buccal mucosa. Palatal lesions most commonly involve the soft palate and uvula in a few cases. Congenital RMS of the lip and tongue has been reported, but lesions of the floor of the mouth are sporadic.[7]In the above case, the lesion involved the right maxilla, including the maxillary sinus, alveolar ridge, and hard palate. It caused destruction of the medial wall of the sinus and extended to the right side, crossing the midline. This extension resulted in deviation of the nasal septum, which clinically presented as nasal congestion.

The differential diagnosis of soft palate masses in children are benign and malignant salivary gland tumors, vascular malformations, hemangiomas, granular cell tumors, hematolymphoid tumors, and metastatic lesions.Although traditionally considered a tumor of skeletal muscle, many RMS originate from sites devoid of musculature. Histologically, tumor cells resemble primitive mesenchyme containing developing or disordered muscle cells and showing a spectrum of differentiation from undifferentiated ‘blastema’ cells to rhabdomyoblasts to terminally differentiated skeletal muscle cells with cross striations. The most primitive rhabdomyoblasts are stellate cells with sparse amphophilic cytoplasm. As rhabdomyoblasts differentiate, they acquire more eosinophilic cytoplasm and become more elongated, forming myotubes and eventually cross-striations. The degree of histopathologic evidence of differentiation varies from case to case. Identification of rhabdomyoblasts may be the key to making a confident diagnosis. Standard immunomarkers include proteins indicative of muscle differentiation, such as desmin and muscle-specific actin, and transcription factors leading to rhabdomyogenic differentiation, such as myogenin and MyoD1. Tumor cells are often diffusely positive for desmin, while myogenin and MyoD1 staining are heterogeneous in different subtypes.[5]

Over the years, the prognosis for rhabdomyosarcoma has significantly improved with a survival rate over 90%. This is based on a combination of therapies, including surgical excision, chemotherapy, and radiotherapy.[8] Interstitial brachytherapy has been tried in recurrent pediatric RMS in orbit, soft palate, gluteal, and anal mass, but there is a dearth of evidence for its use in extremities. Brachytherapy allows the escalation of dose to the target with normal tissue sparing, which makes it a preferred modality for tumors with nearby critical organs.[9]Children with recurrent RMS have a dismal prognosis with long-term survival of less than 15%, particularly if the disease recurs in a metastatic site or area of prior irradiation. The majority of relapses occur within 3 years of therapy completion. Metastatic recurrence is essentially incurable, though treatment may offer palliation. Treatment with surgical resection and adjuvant multi-agent chemotherapy is recommended with new drug combinations such as ifosfamide/ carboplatin/etoposide, docetaxel/gemcitabine, and irinotecan in combination with temozolomide or vinorelbine. Durable remissions for several years may be obtained with aggressive local retreatment and systemic therapy. High-dose chemotherapy followed by hematopoietic stem cell rescue is not advantageous in this group.[1]In the above presented case, although treatment commenced promptly after diagnosis, the child developed severe chemotherapy-related side effects, which led to a deterioration in general health. Despite intensive medical care, the patient unfortunately could not survive the complications and passed away 2.5 months after the initial presentation.

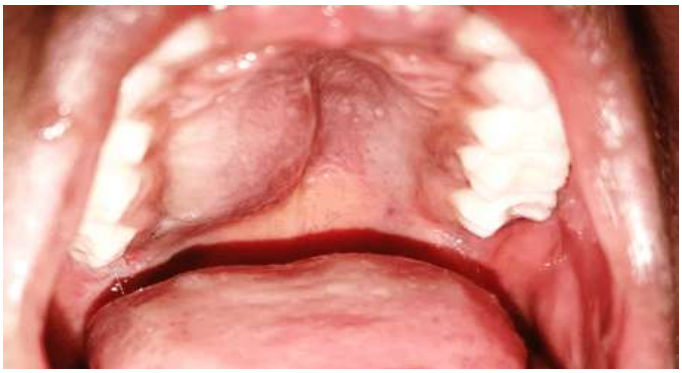


FIGURE. 1. Intraoral aspect showing extensive swelling involving the maxillary alveolus and palate

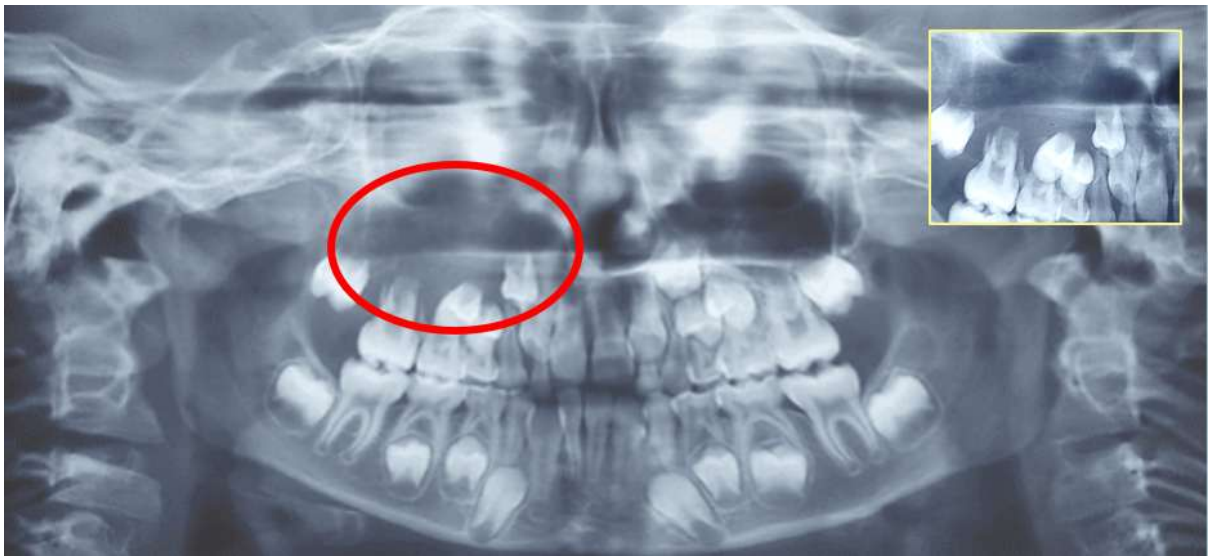


FIGURE. 2. Orthopantomogram showing diffuse lesion involving Right maxilla

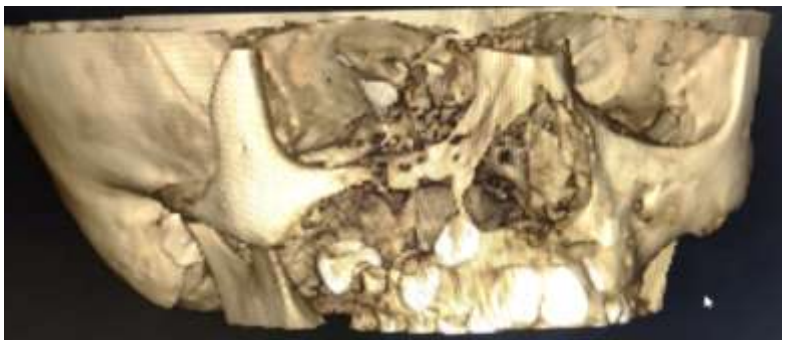
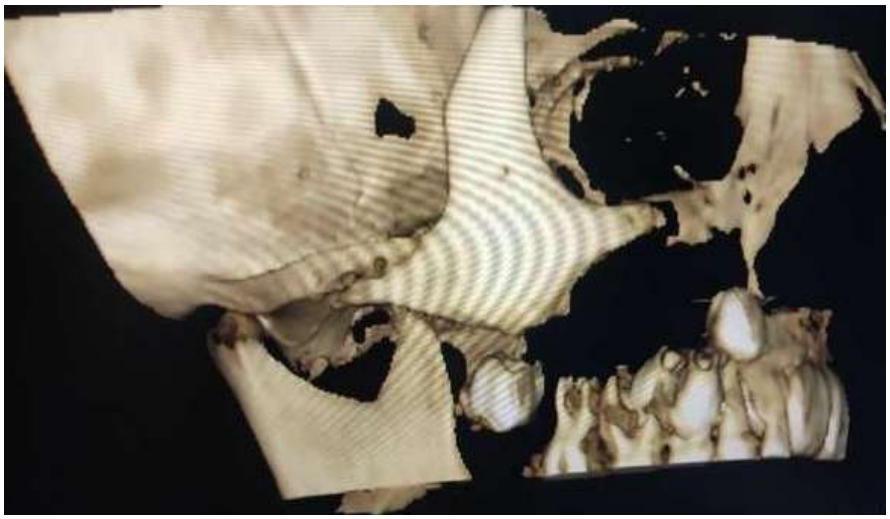


FIGURE.3.a and 3.b:CBCT 3D images showing extensive highly aggressive lesion involving right maxilla suggestive of aggressive benign neoplasm or malignant tumor



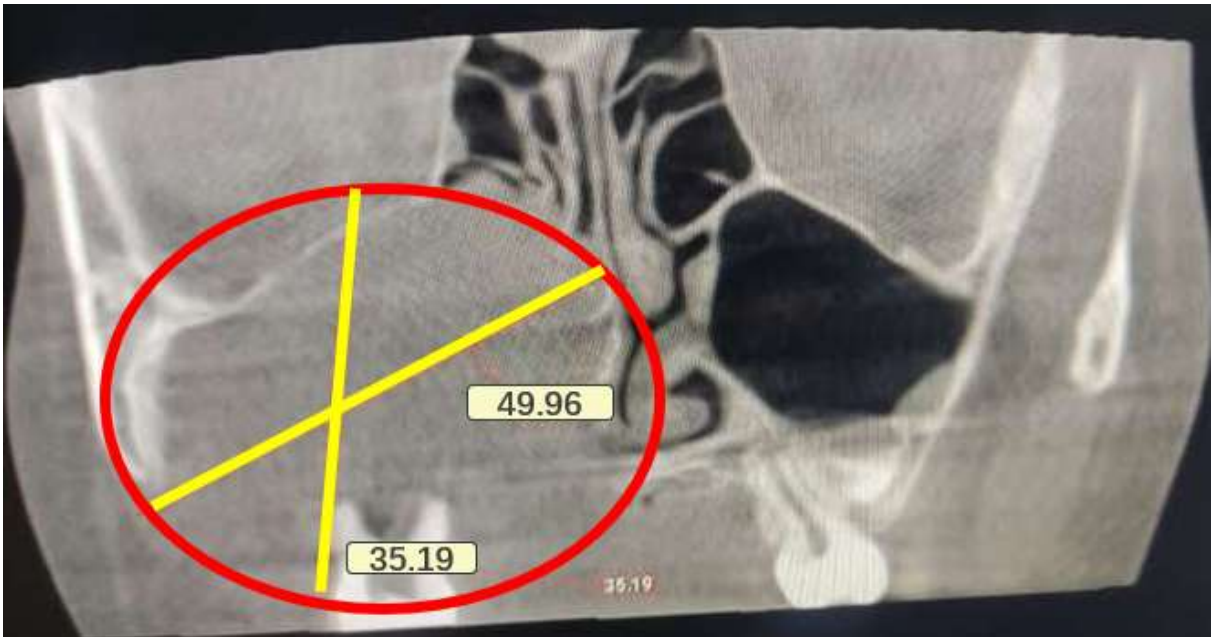


Figure 4. Extensive Expansile Lesion of the Right Maxilla of approximately 46.85 x 49.96 x 35.19 mm in dimension

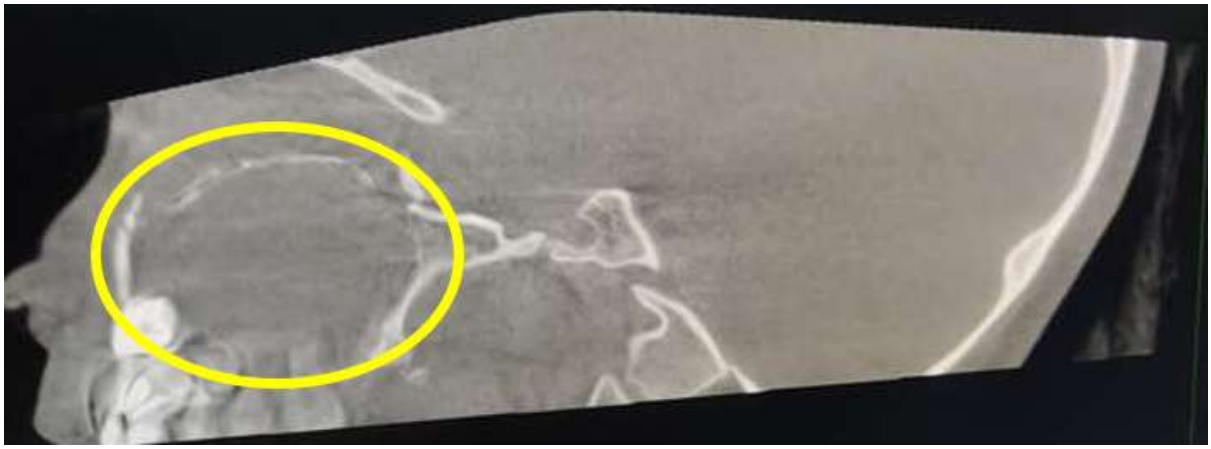


Figure 5. Superior extension of the Lesion pushing the Right Orbital Floor and causing thinning of the Orbit and Superomedially, causing a break in the continuity of the junction of the medial wall and floor of the Right Orbit

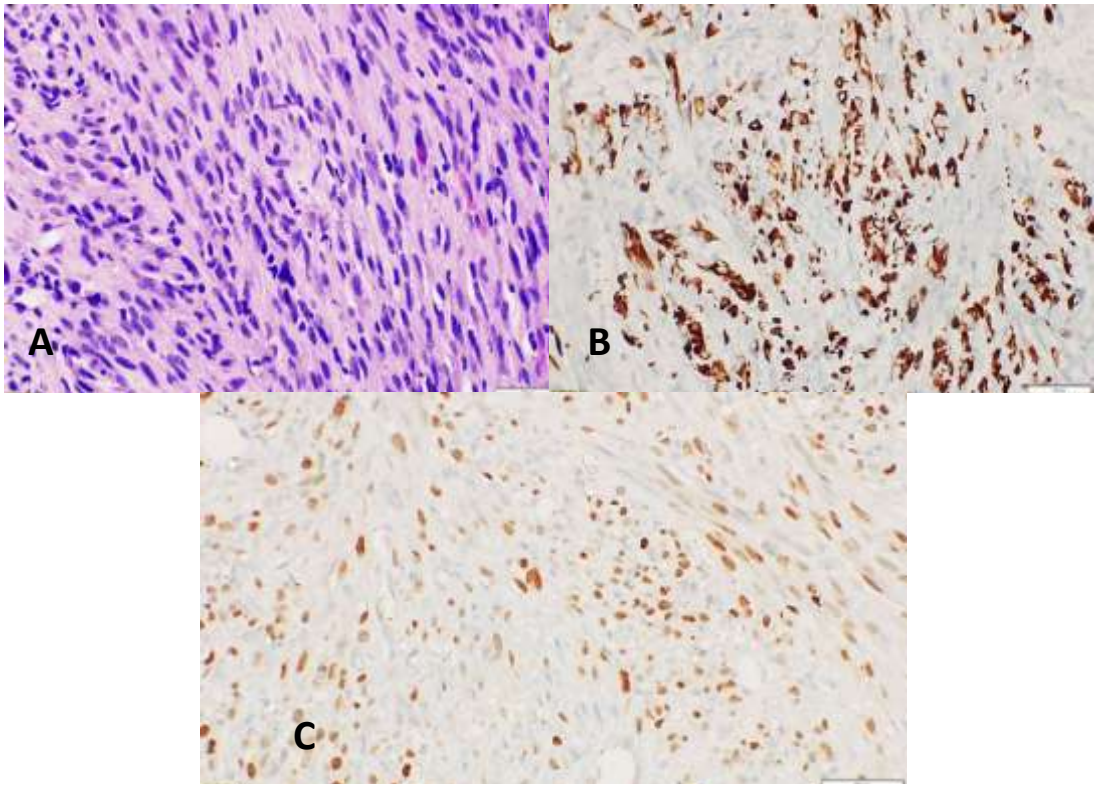


Figure 6.A.H&E 40x: Spindle cells with Ovoid or Elongated 'Cigar-shaped' nuclei. Nuclei are hyperchromatic with scanty cytoplasm. Mitotic figures noted. 6.B.Strong cytoplasmic positivity for Desmin, a marker for skeletal muscle differentiation. 6.C. Nuclear positivity for MyoD1 confirms the skeletal muscle lineage of the neoplasm

Conclusion:-

This case highlights the importance of maintaining a high index of suspicion when evaluating any swelling in children, regardless of its initial presentation. Thorough clinical examination, supported by appropriate radiological and histopathological investigations, is essential for early and accurate diagnosis. Furthermore, regular follow-up and a multidisciplinary treatment approach involving Pediatricians, Oncologists, Surgeons, and dental specialists play a crucial role in improving prognosis and treatment outcomes in such aggressive malignancies.

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