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CASE REPORT

# Ocular Hypertropia Mimicking Inferior Rectus Palsy as an Atypical Presentation of Parameningeal Rhabdomyosarcoma in a Child

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**Introduction:** Rhabdomyosarcoma (RMS) originates from undifferentiated mesenchymal cells that give rise to striated muscles. The symptoms of para-meningeal RMS often resemble those of allergic rhinosinusitis, including nasal congestion, mucus discharge, headache, and occasional nosebleeds. We report a child with atypical clinical presentation of ocular hypertropia secondary to para-meningeal RMS.

**Case Presentation:** A child presented with an upward deviation of the left eye. He had a history of blunt trauma to the face before 5 days. Computed tomography (CT) of the head revealed a soft tissue density involving the left maxillary sinus. The magnetic resonance imaging (MRI) showed a 37.6 mm x 38.4 mm lesion within the left maxillary sinus extending to the orbit, nasal cavity, and premaxillary and retro maxillary areas with a heterogeneous signal and mild heterogeneous enhancements. A biopsy and histopathology confirmed alveolar RMS. The child was treated with chemotherapy and radiotherapy.

**Conclusion:** Pediatric RMS with orbital extension mimicking benign conditions is challenging to diagnose and manage. Hypertropia following eye trauma can obscure severe underlying conditions, such as para-meningeal RMS. The inferior rectus lesion mimicking inferior rectus palsy stresses a thorough evaluation, including imaging and biopsy. Early and accurate diagnosis is crucial for the effective management of children with such aggressive malignancy.

**Keywords:** rhabdomyosarcoma, parameningeal, hypertropia, trauma, case report

## Introduction

Rhabdomyosarcoma (RMS) is a malignancy that originates from the abnormal proliferation of undifferentiated mesenchymal cells that are progenitors of striated muscles. It is the most widely diagnosed soft tissue malignancy among children.[1] Annually, approximately 4.5 per million children are at risk of developing RMS.[2,3] The head and neck are affected sites in 35% of patients with RMS.[2] Based on their location and tendency to invade the central nervous system, RMS of this area are further grouped as orbital, parameningeal, and non-orbital non-parameningeal.[4]

Orbital RMS usually presents as an orbital-occupying lesion, causing either globe dystopia if it arises in the superior nasal quadrant or superior globe displacement if it arises from the orbital floor. However, if it occurs in the sinuses, it has a wide variety of presentations, including nasal obstruction, discharge, sinusitis, local pain, headache, and earache. Advanced cases may develop proptosis, visual disturbances, or cranial nerve palsies.[5]

The nasopharynx, nasal cavity, middle ear, paranasal sinuses, and infratemporal fossa are regions close to the meninges affected by the virulent form of RMS known as parameningeal RMS (PM-RMS).[4,6] It can arise from these parameningeal sites or grow into them from other areas. PM-RMS makes up approximately 20% of RMS cases in children.[7] Thirty percent of PM-RMS cases affect the nasal cavity and nasopharynx, while >60% involve the paranasal sinuses.[3]

Unlike primary orbital RMS, which is typically diagnosed early owing to its distinct symptoms of unilateral, rapidly progressing proptosis, PM-RMS is more challenging to detect because it is not externally visible.[3,8] PM-RMS symptoms often resemble allergic rhinosinusitis, including nasal congestion, mucus discharge, headache, and occasional epistaxis.[8]

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Metastatic orbital tumors generally present with pain, proptosis, diplopia, ptosis, conjunctival redness, or decreased visual acuity, usually following the diagnosis of the primary tumor; however, in 28% of cases, they may be the first indication of malignancy.[9,10] Thus, careful attention to the atypical presentations of PM-RMS is necessary for early detection and treatment of this disease.

This article describes an atypical presentation of maxillary PM-RMS in a child.

## Case Presentation

A 7-year-old boy was brought to the Emergency Department by his mother. The mother complained that the child's left eye had deviated upward. Five days before the deviation, the boy had blunt trauma from a football striking to the left orbit and face. The mother noticed an upward deviation but did not seek immediate medical consultation. The child did not have any pain, blurring of vision, diplopia, loss of consciousness, nausea or vomiting, seizures, or abnormal behavior at presentation.

The child was playful during the ophthalmological examination and had no abnormal head position. His vision was 20/20 in both eyes. He had mild limitation of depression in the left eye but had full extraocular motility in other cardinal directions. In the primary position, hypertropia was noted in the left eye, and it increased in the left gaze with a right-side head tilt, consistent with inferior rectus muscle palsy (Figure 1). He had normal intraocular pressure (IOP) in RE (17mmHg) and LE (16mmHg) as measured by a tonometer (iCare, Vantaa, Finland). No mass was felt around his orbit on palpation, nor was any optic nerve function deterioration noted. Fundus examination showed a healthy optic nerve head and normal retina without brakes. The exophthalmometer measurements at the base (91mm), RE (14 mm), and LE (15 mm) were of normal size.

Computed tomography (CT) of the head (Figure 2) revealed soft tissue density involving the left maxillary sinus extending into the left nasal cavity and left extraconal orbit. This mass could be attributed to an organizing hematoma and was reported to correlate with the clinical findings. The ear, nose, and throat (ENT) review revealed no nasal discharge or obstruction. The left middle turbinate was congested with purulent discharge adherent to the mucous membrane. The maxillary opacification was attributed secondary to trauma, but an urgent Magnetic resonance imaging (MRI) sinus with contrast was recommended to rule out other causes.

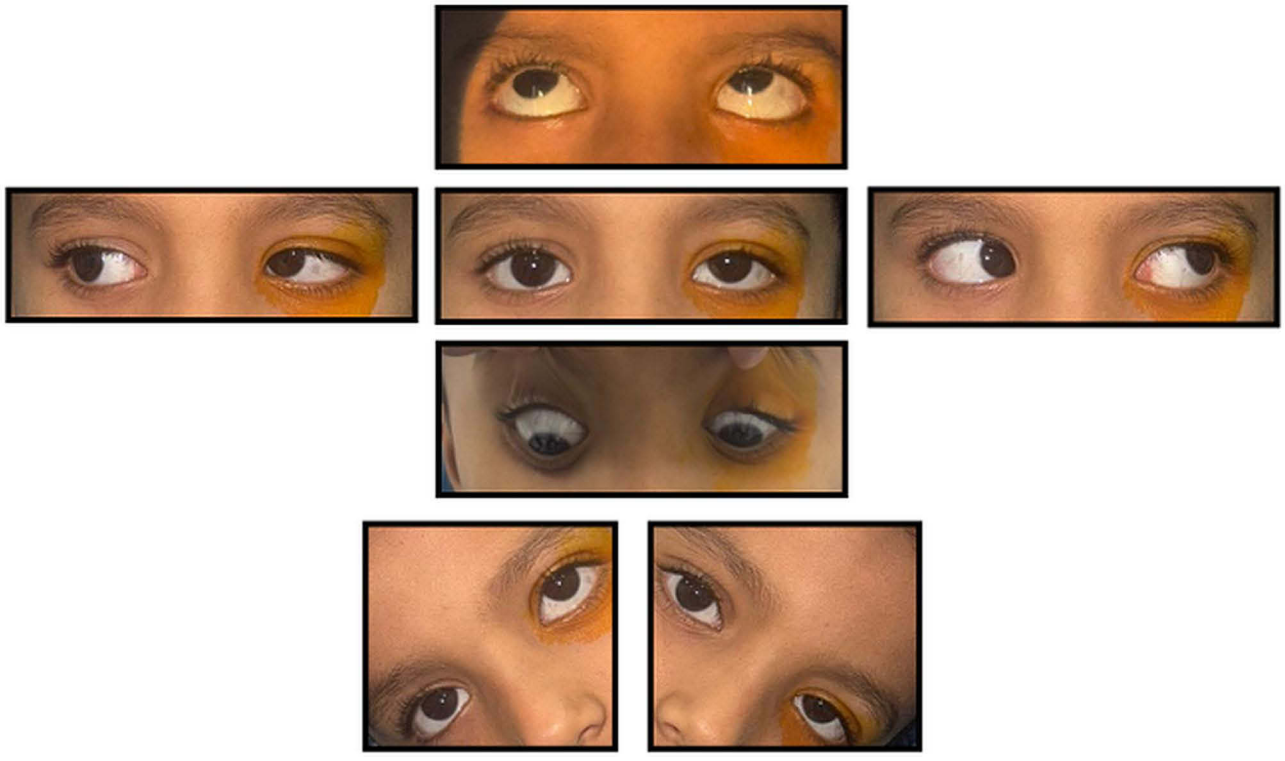

**Figure 1** Position of both eyes in different gaze noted after blunt trauma in a child with para-meningeal rhabdomyosarcoma. In primary gaze, there is left eye hypertropia. This worsens on the left gaze and the right head tilt. There is a −1 degree limitation on depression on infra-duction. The yellow discoloration was due to fluorescein stain, which was used to detect any epithelial defect.

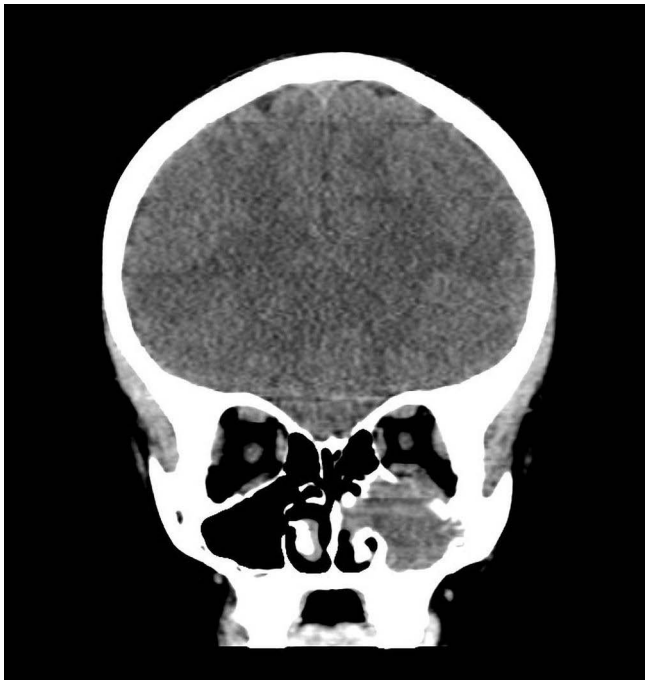

**Figure 2** Coronal cuts of computerized tomography image of a child with para-meningeal rhabdomyosarcoma. The star shows a soft tissue shadow in the left maxillary sinus extending into the left nasal cavity and the left extra conal orbit.

The Magnetic Resonance Imaging (MRI) showed a 37.6×38.4 mm lesion within the left maxillary sinus, extending to the orbit, nasal cavity, premaxillary, and retro-maxillary areas with a heterogeneous signal and mild heterogeneous enhancement. There was also an underlying abnormal marrow signal in the left posterior maxillary alveolus with abnormal thickening and signal in the gingiva, which was suspected to be a neoplastic process (Figure 3).

A biopsy of the left maxillary sinus and left nasal cavity showed malignant round cell neoplasm positive for desmin, myogenin, and myogenic differentiation 1. Fluorescence in situ hybridization was positive for FOXO1.

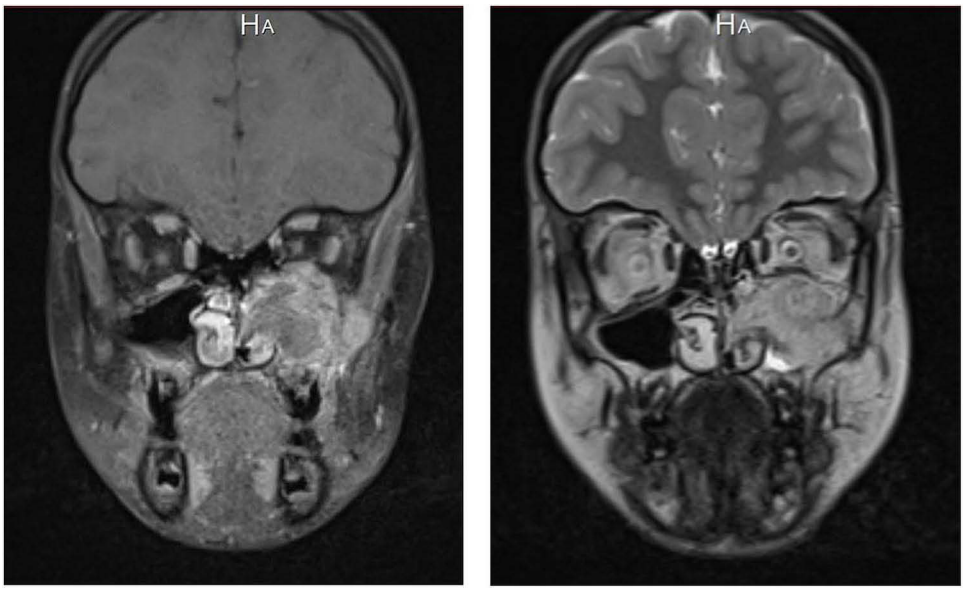


**Figure 3** Magnetic Resonance Images of the skull and orbits of a child with para-meningeal rhabdomyosarcoma. The right image is coronal cut T1 weighted and fat suppression with contrast. It shows a heterogeneous enhancing lesion of 37.6 mm x 38.4 mm in the left maxillary sinus isotense to the extraocular muscles, invading the left inferior rectus muscle and infraorbital fat. The left image is a coronal cut T2 weighted with contrast, showing the hyperintensity of the lesion to the extraocular muscles and enhancement.


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**Figure 4** The child's left eye with para-meningeal rhabdomyosarcoma ten days after initial presentation. The eye has lagophthalmos and severe conjunctival chemosis.

Based on these clinical and imaging findings, the patient was diagnosed to suffer from alveolar fusion-positive RMS. A full metastatic workup included cerebrospinal fluid analysis, bone marrow aspiration and biopsy, chest CT, brain MRI, bone scan, and positron emission tomography. It did not reveal any metastatic lesions.

One month after the presentation, the child developed severe conjunctival chemosis, high IOP, and lagophthalmos. (Figure 4) He was given thrice daily dorzolamide 2% eye drops to lower the IOP.

The Tumor Board labeled this child to have PM-RMS stage III. According to the TNM classification, the tumor was classified as T2bNxM0, representing intermediate risk based on Children's Oncology Group Rhabdomyosarcoma Risk Stratification.[11] The treatment plan included radiotherapy and chemotherapy. The chemotherapy included vincristine, actinomycin D, and cyclophosphamide (VAC) in high doses. The child received 45 Gy/25 fx using rapidArc technique. Five cycles were given, and treatment is still ongoing. Three months after the start of treatment, there was a marked decrease in the size and extension of the tumor, with the 3 months later image showing the orbits and optic nerve sheath free of tumor. Six months after initiating the therapy, the patient's appearance improved, and his IOP normalized. Therefore, 2% of dorzolamide eye drops were discontinued. The child continues to be under the care of a multidisciplinary team.

A timeline of events is given in Table 1 for a better understanding.

**Table 1** Timeline for Events in the Care of a Child with Inferior Rectus Palsy and Para Meningeal Rhabdomyosarcoma

| | Event | Date |
|---|---|---|
| 1 | Blunt trauma of the face and eye | 02/02/2024 |
| 2 | The child was brought to the eye emergency department | 07/02/2024 |
| 3 | Clinical assessment and workup for ocular palsy | 07/02/2024 |
| 4 | Computerized tomography of head and orbit | 07/02/2024 |
| 5 | Seen by the ENT team and their impression of maxillary opacification for urgent MRI sinus with contrast | 08/02/2024 |
| 6 | Magnetic resonance imaging of the face, sinuses, and orbits | 21/02/2024 |
| 7 | Biopsy of the tumor mass | 03/03/2024 |
| 8 | Presentation with lower lid chemosis | 07/03/2024 |
| 9 | Diagnosis and Evaluation for metastasis | 10/03/2024 |
| 10 | Chemotherapy | 14/03/2024 -ongoing, |
| 11 | Radiotherapy | 21/3/2024 to 26/5/24 |
| 12 | Last follow-up and ophthalmic evaluation | 5/8/2024 |

## Discussion

We report an uncommon presentation of hypertropia as an early manifestation of PM-RMS. This can easily be mistaken for other conditions, particularly after trauma. This is the case of hypertropia as a presenting sign of PM-RMS caused by inferior rectus infiltration and was accidentally noted following physical trauma to the child. Trauma is known to cause local inflammation, which releases proinflammatory mediators at the site of tissue trauma, producing conditions favorable for dormant tumor cell proliferation and metastasis.[12] However, a five-day gap between trauma and presentation does not seem to be a trigger for the tumor extension. This can explain the rapid progression of ophthalmic symptoms, such as hypertropia, proptosis, decreased vision, and increased IOP. These signs indicated orbital extension.

Similar to our case, Shahidatul-adha M. et al reported three children having different ocular presentations: one experienced sudden vision loss, another had unilateral eye protrusion without visual issues, and the third had a sudden onset of left divergent squint leading to progressive blurry vision and painless proptosis.[13] Despite being labeled as symptomatic, PM-RMS is aggressive and frequently spreads locally and intracranially, with rates of meningeal spread reported between 26% and 35% in the literature.[3,8,14]

Consequently, diagnostic imaging is essential for evaluating patients who present with ocular deviation following trauma. Ocular deviation after trauma can indicate severe underlying conditions, such as orbital fractures, intracranial hemorrhages, or tumors. CT and MRI are vital for revealing these pathologies.[15] MRI is the preferred imaging modality for evaluating the extent of the primary tumor, which helps define tumor boundaries and the involvement of adjacent structures.[16] Although imaging provides detailed anatomical information, a definitive diagnosis of RMS requires histopathological examination.[17]

The management of this case involved a multidisciplinary team, including oncology, ophthalmology, ENT, and radiology specialists. The patient was treated according to the intermediate-risk protocol of the high-dose VAC regimen, as recommended for pediatric RMS with para-meningeal involvement. VAC has been the standard of care for years and continues to be the best option, as studies trying novel agents have not demonstrated benefits.[18] The treatment of such cases is multimodal. Radiotherapy, like surgery, is used for local control, while chemotherapy is used for presumed micro-metastases.[19,20]

## Conclusion

This case emphasizes the complexity of diagnosing and managing pediatric RMS with orbital extension, particularly when the initial symptoms mimic benign conditions. Hypertropia following eye trauma can obscure severe underlying conditions, such as PM-RMS. The initial misdiagnosis of inferior rectus palsy underscores the importance of a thorough diagnostic evaluation, including imaging and biopsy. Early and accurate diagnosis is crucial for effective management and improvement of outcomes in children with this aggressive malignancy.

## Ethics Approval and Informed Consent

We have obtained informed consent from the patient's parents to publish this case report and any accompanying images. No institutional approval is required to publish case details.

## Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis, and interpretation, or all these areas; took part in drafting, revising, or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

## Disclosure

The authors report no conflicts of interest in this work.

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