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Case Report

Wide-Excision Choice in Orbital Rhabdomyosarcoma on an 8-Year-Old Patient in a Low-Resource Setting: A Case Report

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Keywords

Rhabdomyosarcoma · Embryonal rhabdomyosarcoma · Orbital rhabdomyosarcoma · Pediatric tumor

Abstract

Introduction: Rhabdomyosarcoma is the most common neoplasm of skeletal myoblast-like cells in children aged 5–8 years. It typically occurs in the head, neck region, genitourinary tract, retroperitoneum, and extremities. ***Case Presentation:*** An 8-year-old boy complained of a lump under his left eye that he had felt for the past 4 years. Initially small, it gradually grew larger although there were no accompanying symptoms of redness, pain, itching, or eye movement disorders, and no history of trauma. Visual acuity in the left eye was measured at 6/15, with pinhole correction improving to 6/6, while the right eye was 6/6. Intraocular pressure in both eyes was 15 mm Hg. Anterior segment examination of the left eye revealed a mass on the lower eyelid with hard consistency, no erythema, no tenderness, with a flat surface, immobility, and all aspects of the conjunctiva, cornea, pupil, and lens appearing normal. A contrast computed tomography scan showed a solid mass measuring 2.1 × 1.9 × 1.9 cm, suggesting a left inferior eyelid mass. The patient underwent surgical excision of the tumor under general anesthesia. Histopathological examination confirmed the diagnosis of embryonal rhabdomyosarcoma. The patient was scheduled for chemotherapy. ***Conclusions:*** Orbital rhabdomyosarcoma typically presents with sudden onset and rapid proptosis, without a history of trauma or respiratory tract infections. The embryonal subtype is the most common and has a better prognosis with combined therapy including surgery, chemotherapy, and radiation. In low-resource settings, early clinical suspicion and thorough physical examination are vital, as limited access to medical tools can complicate

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management. Treatment should be adapted based on available resources, and regular follow-up is essential to monitor outcomes and ensure optimal care. Any swelling in children should be carefully examined for early detection and effective intervention.

Introduction

Rhabdomyosarcoma is a rare connective tissue cancer, with a global incidence rate of 4.3 per million population. Nevertheless, it is the most common neoplasm of skeletal myoblast-like cells in children aged 5–8 years. Approximately half of all cases manifest within the first decade of life, with a gender predilection toward males. It typically occurs in the head, neck region, genitourinary tract, retroperitoneum, and extremities [1, 2].

Rhabdomyosarcoma in the orbital regions often presents with rapid-onset proptosis, chemosis, ophthalmoplegia, and painful ocular movements. The rate of progression varies; however, acute or even overnight progression has been described in many cases of orbital rhabdomyosarcoma [2]. Given the clinical characteristics, it is not uncommon for clinicians to mistake it with inflammatory conditions, such as orbital cellulitis and myositis [3].

Prompt diagnosis is of paramount importance to prevent the disease from spreading and threatening patients' vision and life. If left untreated, both local and systemic metastases can occur [1, 2]. In light of the critical importance of prompt diagnosis and treatment, we present a case study involving an 8-year-old patient with orbital rhabdomyosarcoma of the embryonal type in a low-resource setting. This case underscores the challenges faced in diagnosing and managing this condition, particularly in resource-constrained environments.

This case has been reported in line with the 2013 CAse REport (CARE) criteria [4]. The CARE Checklist has been completed by the author for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000541645).

Case Presentation

An 8-year-old male presented with a chief complaint of a progressively enlarging lump inferior to the left inferior palpebra, which he had noticed for the past 4 years (Fig. 1a). He was referred from a small hospital near his hometown, located more than 400 km from our tertiary healthcare facility. The initial size of the lump was small, but it had gradually increased in size over time. Despite its growth, the patient reported no accompanying symptoms such as redness, pain, itching, or disturbances in eye movement. Furthermore, there was no history of trauma to the area.

Upon examination, visual acuity in the left eye was measured at 6/15, with improvement to 6/6 through pinhole correction, while the right eye maintained a visual acuity of 6/6. Eye movements were normal in all directions. Intraocular pressure in both eyes was within normal limits, measured at 16 and 17 mm Hg for the right and left eyes, respectively. Anterior segment examination of the left eye revealed a palpable mass on the lower eyelid characterized by its hard consistency. No erythema or tenderness was noted, and the surface of the mass appeared flat with immobility upon palpation. Notably, all aspects of the conjunctiva, cornea, pupil, and lens appeared normal with intact light reflex. There was a discordance of margin reflex distances (MRDs) and vertical palpebral fissures (VPFs) in the left eye (MRD1 +3, MRD2 +5, and VPF 8 mm) compared to the right (MRD1 +4, MRD2 +6, and VPF 10 mm).

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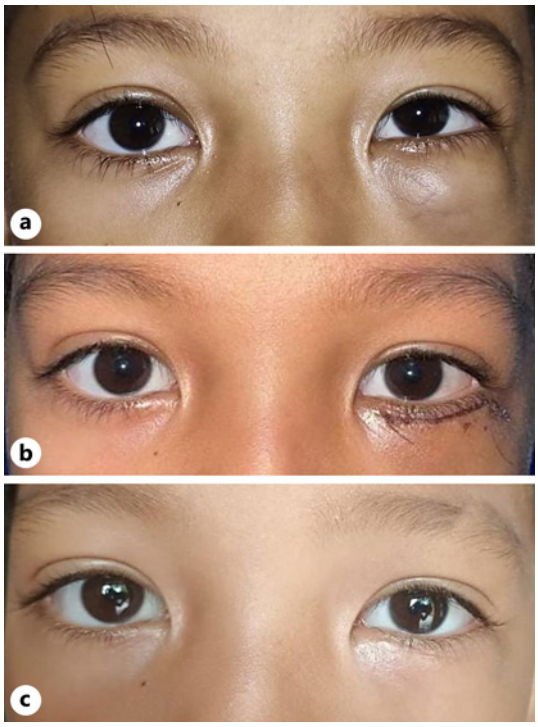

Fig. 1. a Patient's initial eye condition. **b** Patient's eye condition 1 week after surgery. **c** Patient's current eye condition.

Further investigation through contrast-enhanced computed tomography (CT) imaging revealed a solid mass measuring 2.1 × 1.9 × 1.9 cm, localized to the left maxillary sinus, infiltrating the left orbital bone (Fig. 2). Based on these findings, a diagnosis of embryonal rhabdomyosarcoma in the inferior part of the eyelid was suspected. Afterward, the patient underwent surgical excision of the tumor under general anesthesia.

We managed to excise a 2 × 1.5 × 0.5 cm tissue (Fig. 3) for further testing by the Department of Anatomic Pathology. Histopathological examination of the excised tissue confirmed the presence of embryonal rhabdomyosarcoma. The tissue sample from the tumor of the inferior palpebra of the left eye shows proliferation of spindle-shaped nuclei cells, atypical in appearance, arranged in interlacing fascicles. There are cells with elongated eosinophilic cytoplasm and ganglion-like cells with eosinophilic cytoplasm (rhabdoid cells) infiltrating between fatty tissue and perineural tissue. In light of this diagnosis, the patient was scheduled to undergo prompt chemotherapy as part of the treatment plan.

After the surgery, the patient's condition was found to be good. Initial follow-up revealed normal eye function with minimal swelling in the left eye (Fig. 1b). Nevertheless, due to the considerable distance between the patient's hometown and our healthcare facility (400–500 km), treatment compliance has been found to be poor. The patient comes from a low-socioeconomic background and faces difficulties in returning to our hospital. However, the patient's mother reports that her son has not experienced any further swelling since the surgery up to the time of this report (Fig. 1c).

Discussion

Orbital rhabdomyosarcoma is a rare but aggressive malignancy, most commonly affecting children aged 5–8 years. It typically presents with rapid-onset proptosis, chemosis, ophthalmoplegia, and painful ocular movements. This typical clinical manifestations of orbital

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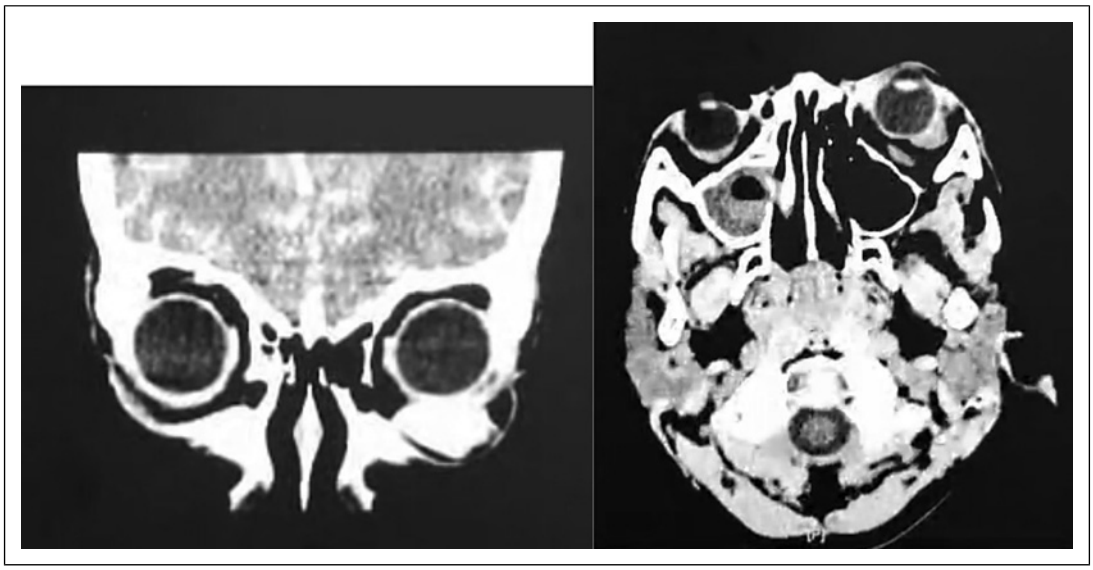

Fig. 2. CT-scan result of the patient.

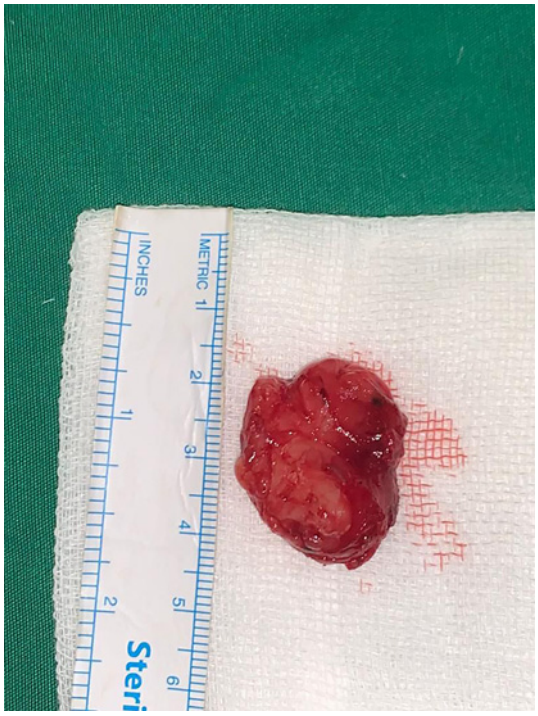

Fig. 3. Extracted mass of the tumor.

rhabdomyosarcoma, presented as a painful rapidly progressing 2 × 2 cm mass within 1 month in a 9-year-old female patient, was reported in a case report by Thien et al. [5]. Moreover, Shrestha et al. [6] also reported a case of rapidly progressing rhabdomyosarcoma, initially presented as protrusion of the right eye within 3 months in a 14-year-old male patient. However, in our case, the 8-year-old boy presented with a gradually enlarging lump under his left eye, consistent with the typical presentation of orbital rhabdomyosarcoma [1, 2]. Despite the absence of overt symptoms such as redness or pain, the persistent growth of the mass

over 4 years raised concerns, prompting thorough clinical evaluation. CT scan unveiled a solid mass in the left inferior eyelid, aligning with suspicions of a malignant process. Surgical excision of the tumor followed by histopathological examination confirmed the diagnosis of embryonal rhabdomyosarcoma.

Orbital rhabdomyosarcoma is often misdiagnosed as an inflammatory response due to infections, such as orbital cellulitis and myositis. This is to be expected, considering rhabdomyosarcoma often occurs in the genitourinary tracts (22% of all cases) and extremities (18%), compared to the orbits (10%) [3, 7, 8]. In our case, although other diagnoses were considered, the lack of fever and local inflammatory responses pointed to a tumor origin. Further CT scans and histopathological tests confirmed our suspicion.

Our case is unique. In contrast to typical cases of orbital rhabdomyosarcoma, which often present with rapidly developing proptosis (90% of all cases), globe displacement (80%), and blepharoptosis (40%), our case involved an 8-year-old boy with a gradual enlargement of a lower eyelid mass over 4 years. This slow progression without proptosis or displacement is not typical. Additionally, the mass was located in the lower eyelid, whereas the usual predilection sites of rhabdomyosarcoma are in the superior and superonasal areas. This difference in location prevented significant proptosis [3, 7, 8]. These contrasting features underscore the unique and atypical presentation of orbital rhabdomyosarcoma in this case, emphasizing the need for thorough evaluation even in the absence of classic symptoms.

Given the successful follow-up after surgery, we can safely predict a good prognosis for the patient. Improvements in adjuvant therapy, such as chemotherapy and radiotherapy, have significantly increased the overall survival rate for rhabdomyosarcoma patients, reaching up to 90% compared to the previous rates of around 25–30% during the 1960s [2]. The American Academy of Ophthalmology recommends comprehensive ocular examinations every 3–4 months during the first year following treatment. These exams should include assessments of best-corrected visual acuity, external ocular health for signs of dry eye, proptosis, masses, and motility disturbances, slit-lamp biomicroscopy for cataract detection, ophthalmoscopy to check for radiation retinopathy, and orbital CT or MRI scans to evaluate for any residual or recurrent tumors. In the second year, 4- to 6-month follow-up should continue for several years. Additional adjuvant therapy would be administered during these follow-up sessions [8].

However, treatment compliance has been challenging due to distance challenges. The patient, coming from a low-socioeconomic background, struggles to return for follow-up visits. Although the Indonesian government provides a universal healthcare program (*Jaminan Kesehatan Nasional*; JKN) to cover medical expenses, adequate healthcare delivery still faces indirect financial obstacles [9]. In our particular case, the lack of accessible and affordable means of transport is the biggest hurdle to completing follow-up.

Despite showing favorable findings post-therapy, embryonal orbital rhabdomyosarcoma can still exhibit aggressive behavior with distant metastases [10]. This highlights the importance of genetic and molecular diagnosis in guiding treatment decisions and prognostication. Molecular assays, such as detecting the PAX-FOXO1 fusion transcript, are essential for accurately identifying rhabdomyosarcoma subtypes and predicting their clinical behavior [1, 11]. Nevertheless, our center did not have access to such sophisticated molecular assays for precise diagnosis and prognostication, making wide-excision surgery the mainstay of management.

With significant problems in ensuring follow-up compliance and access to advanced methods of diagnosis in low-resource settings, primary care clinicians should have the necessary knowledge to correctly determine tumor origin in every case of children with chronic swelling and to recognize signs of tumor relapse. Early diagnosis could significantly help prompt surgical treatment, with wide-excision being the best available

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management to prevent further recurrence in such situations. Our case underscores the importance of tailored approaches to diagnosis and treatment in resource-limited settings, highlighting the need for vigilant strategies to improve access to comprehensive cancer care.

Conclusion

It is important to approach any swelling in children with thorough examination and vigilant follow-up, particularly in low-resource settings where access to comprehensive care may be limited. Our case of orbital rhabdomyosarcoma underscores the importance of early diagnosis and multidisciplinary management. Utilizing a combination of excisional surgery, chemotherapy, and radiation therapy has significantly improved the prognosis for patients with this aggressive malignancy. Although often times not available due to socioeconomic challenges, continuous monitoring and adherence to treatment protocols are crucial to ensure optimal outcomes. As such, enhancing awareness and access to comprehensive care remain paramount in achieving successful outcomes for children afflicted with orbital rhabdomyosarcoma and other similar conditions.

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Statement of Ethics

This study, being a case report, did not require formal ethical approval as per the policies of the Institutional Review Board at the Prof. R. D. Kandou General Hospital-Faculty of Medicine, Sam Ratulangi University. Nevertheless, informed consent was obtained from the patient, and their confidentiality and privacy were strictly maintained throughout the study.

Written informed consent was obtained from the patient's parents/legal guardian for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Conflict of Interest Statement

The author declares that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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Author Contributions

Conceptualization, methodology, validation, formal analysis, investigation, resources, data curation, writing – original draft preparation, writing – review and editing, and project administration: V.S. The author attests that they meet the current ICMJE criteria for Authorship.

Data Availability Statement

The data that support the findings of this study are not publicly available due to their containing information that could compromise the privacy of research participants but are available from the corresponding author (V.S.) upon reasonable request.

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