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Case Report

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Challenges in primary orbital Ewing sarcoma: A case report

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Abstract

Primary Orbital Ewing Sarcoma (POES) is an exceptionally uncommon cancer. We present a case study of POES in a young woman that highlights the intricate diagnostic process, therapeutic interventions, and the essential role of a multidisciplinary healthcare team. Despite the patient's parents' decision to seek alternative approaches, delaying treatment, a comprehensive approach resulted in a favorable outcome, emphasizing the importance of early detection, and coordinated care. This case report adds to the restricted information available on this rare clinical entity, providing valuable insights into its diagnosis and management.

Introduction

Ewing sarcoma (EWS) is a rare and aggressive form of primary bone cancer that typically affects children, adolescents, and young adults. The tumor was initially identified as diffuse endothelioma of bone by James Ewing [1]. It is classified as a member of the Ewing Sarcoma Family of Tumors (ESFTs). Primary Ewing sarcoma of the head and neck region typically affects the mandible or maxilla and is classified as a small, round-cell neoplasm of neuroectodermal origin [2]. POES is a subtype of EWS that originates in the orbit of the eye and is extremely infrequent [3].

EWS is a rare type of childhood cancer, making up only 2% of cases. It is the second most common malignant bone tumor in young adults and children, with osteosarcoma being the first [4]. This cancer usually develops in the long bones such as the arms, legs, or pelvis, but it can also be found in soft tissues. EWS is more commonly found in males during the second decade of life [5]. POES accounts for only a small portion of EWS cases [6-11].

Common symptoms of EWS include localized pain, swelling, and, in some instances, the presence of a palpable mass at the tumor site. As the disease advances, patients may also present symptoms such as fatigue, fever, and unintended weight loss [12]. Common laboratory findings include anemia, elevated Erythrocyte Sedimentation Rate (ESR), and heightened levels of Lactate Dehydrogenase (LDH) [13]. Furthermore, EWS carries the risk of metastasis to distant regions of the body, with the lungs and bones being the most frequent sites of secondary involvement [5]. Patients with POES may exhibit various symptoms, including proptosis, limited eye movement, and vision impairment. Notably, the clinical presentation frequently resembles that of other orbital tumors, which can complicate the diagnostic process [11].

Diagnosis of POES is established through a two-step process. Initially, a combination of imaging studies, such as orbital magnetic resonance imaging (MRI) and computed tomography (CT) scans, is employed to assess the extent and characteristics of the orbital mass [14]. Subsequently, for a definitive diagnosis, a histopathological examination of a biopsy specimen is crucial. This examination reveals specific features, including the presence of small, round, blue cells, and the expression of distinctive immunohistochemical markers such as CD99 and FLI-1 [15,16].

Effective management of POES requires a multidisciplinary

approach, uniting the expertise of oncologists, ophthalmologists, and radiation therapists. Treatment strategies encompass a combination of therapies, often beginning with neoadjuvant chemotherapy protocols with agents such as vincristine, doxorubicin, cyclophosphamide, and ifosfamide [17]. Exenteration is considered to remove the tumor and addressing metastatic lesions when viable [18,19]. Following surgery, adjuvant radiotherapy may be employed to further minimize the risk of local recurrence [20].

The prognosis for POES is multifactorial and hinges on several key factors, including the stage of the tumor, the patient's response to treatment, and the extent of surgical resection [21]. The prognosis for patients with recurrent EWS is very poor, with a 5-year survival rate of 13% [22]. Patients with relapsed EWS also have an unfavorable prognosis [23]. The overall 5-year survival rate for localized Ewing tumors is 70%, while metastatic EWS has a 5-year survival rate of 30%. Long-term follow-up is imperative to diligently monitor for any late effects resulting from treatment and the potential for disease recurrence. It's essential to acknowledge that the rarity of this condition translates to a limited pool of data available for a comprehensive assessment of prognosis, underscoring the significance of ongoing research and clinical observation in refining our understanding of the disease's outcomes [18].

Case presentation

A 12-year-old previously healthy woman presented with a three-week history of progressive proptosis, and pain in her right eye. On examination, she had moderate right-eye proptosis and extraocular movements were preserved (Figure 1a). Fundus examination revealed optic nerve edema on the same eye. There were no palpable masses or lymphadenopathy. Initial CT was performed, the scan demonstrated a mass centered within the right orbital apex, infiltrating the adjacent structures (Figure 1b). A core needle biopsy was performed, and histopathological examination revealed small, round, blue cells with focal rosette formation, consistent with EWS. Immunohistochemistry further confirmed the diagnosis with positive staining for NKX2.2 (Figure 2a-b).

Unfortunately, the parents decided to pursue a different treatment approach, causing a one-month delay in starting the recommended treatment protocol. Nevertheless, as the condition failed to improve and instead worsened, they ultimately opted to follow the original plan. Upon further examination, the patient exhibited marked right-eye proptosis, pain, limited extraocular movements and a decrease visual acuity to negative perception of light (Figure 1c). An additional MRI was performed it demonstrated a nonhomogeneous mass within the right orbital apex, infiltrating the adjacent structures with severe proptosis (Figure 1d). Subsequent systemic evaluation, including bone scintigraphy, CT of the chest, abdomen, and pelvis, and bone marrow aspiration, showed no evidence of metastasis, thus confirming the diagnosis of POES.

Neoadjuvant chemotherapy was initiated with a regimen comprising vincristine, doxorubicin, and cyclophosphamide (VAC). A right exenteration procedure was conducted (Figure 3a), and the postoperative histopathological analysis revealed that the surgical margins were free from malignancy. Adjuvant radiotherapy was administered to the surgical site to minimize the risk of local recurrence. The patient underwent a total of six cycles of adjuvant chemotherapy. Serial imaging showed no evidence of local recurrence or distant metastasis. An external orbital prosthesis was fitted onto the right side (Figure 3b) and patient continues to receive regular follow-up care to monitor for potential late effects of treatment and recurrence.

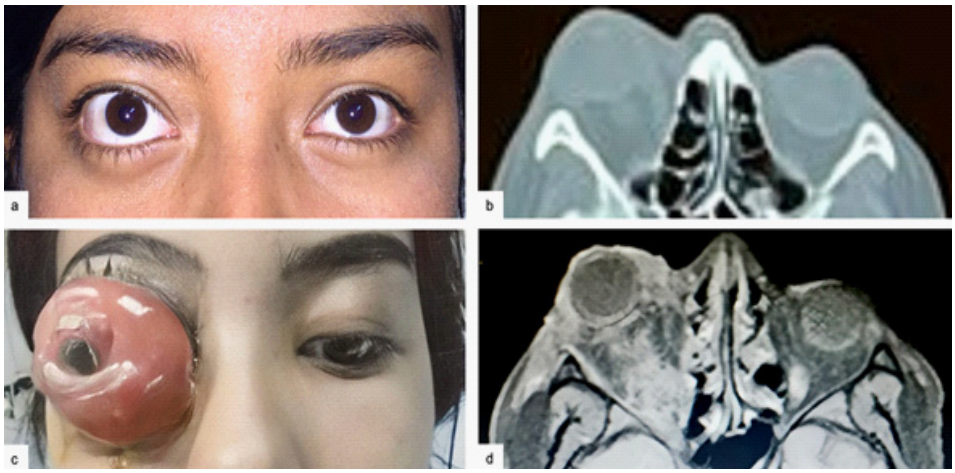

Figure 1: (a) Right eye exhibits a subtle proptosis compared to the left eye. **(b)** Orbita CT showed a mass centered within the right orbital apex, infiltrating the adjacent structures. **(c)** Severe right proptosis accompanied by purulent discharge and marked chemosis. **(d)** Orbita MRI revealed a nonhomogeneous mass within the right orbital apex, infiltrating the adjacent structures with severe proptosis.

Discussion

The case of this young patient with POES underscores several significant aspects of this rare disease. POES often mimics other orbital tumors such as orbital hemangioma, orbital cellulitis, Graves' ophthalmopathy, orbital lymphoma, orbital dermoid cyst, orbital metastasis, orbital neurofibroma, cavernous hemangioma, venous malformation, rhabdomyosarcoma, idiopathic orbital inflammatory syndrome, optic nerve glioma, and orbital mucocele [24]. Therefore, a high index of suspicion for this entity is needed, especially in young individuals with atypical orbital masses.

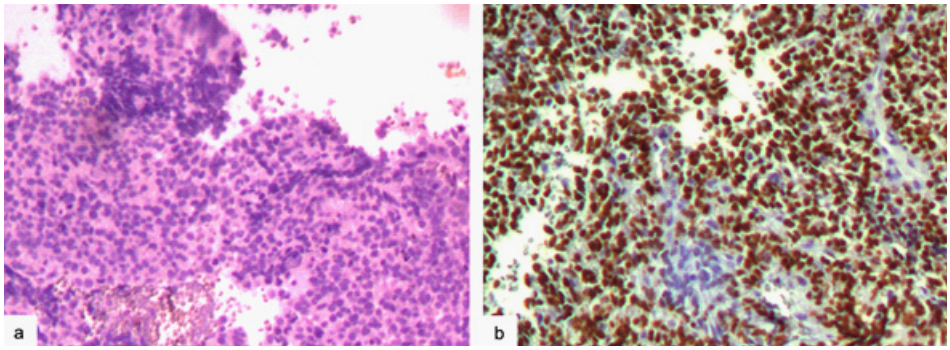

Figure 2: (a) Clusters of small, round cells characterized by limited cytoplasm and the presence of mitotic figures, Indistinct cytoplasmic membranes (hematoxylin and eosin stain). **(b)** Strong diffuse nuclear staining for NKX2.2.

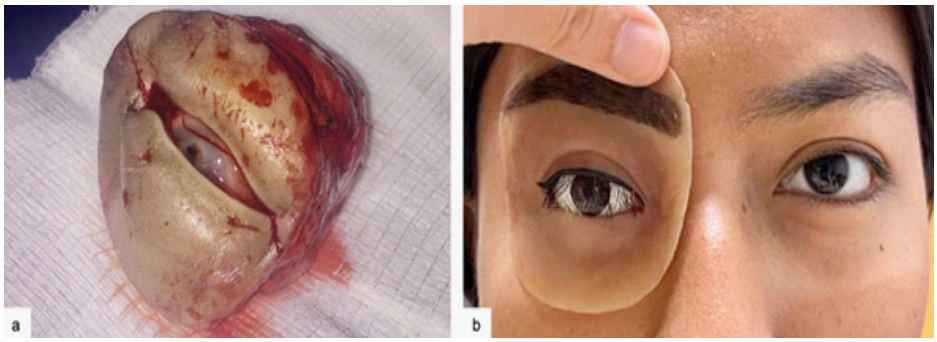

Figure 3: (a) Right exenteration. **(b)** Right external orbital prosthesis.

Early diagnosis is crucial in the management of POES. In our case, the patient's parents' decision to discontinue treatment serves as a striking reminder of the grave repercussions that may arise when diagnosis is postponed or when patients are not well-informed about the criticality of adhering to their treatment regimen. Providing psychological support and addressing the patient's concerns and fears can help ensure treatment fulfillment.

Multidisciplinary collaboration is another crucial aspect highlighted in this case. Managing POES requires the expertise of oncologists, ophthalmologists, and radiation therapists working cohesively to determine the most appropriate treatment strategy. A well approach was initiated in this case but ultimately interrupted. However, treatment was initiated, resulting in the most favorable outcome.

Conclusions

In this case, of a patient with POES who ceased treatment serves as a poignant reminder of the challenges associated with this rare malignancy. Successful management of this condition relies on a collaborative approach, early diagnosis, and aggressive treatment strategies. A diligent long-term follow-up are imperative for achieving favorable outcomes in these uncommon cases. Enhancing healthcare providers' awareness of this distinctive form of EWS can lead to improved recognition and more effective management. Lastly, ensuring patient education and support is essential to ensure treatment compliance and ultimately improve the prognosis for this aggressive disease.

References

1. The classic. Diffuse endothelioma of bone. By James Ewing. 1921. Clin Orthop Relat Res. 1984; (185).
2. Sendul SY, Ucgul C, Kabukcuoglu F, Dirim B, Guven D. Primary orbital Ewing's sarcoma presenting with local recurrence to maxillary sinus shortly after tumor resection. J Surg Case Rep. 2015; 2015(7).
3. Sbaraglia M, Righi A, Gambarotti M, Dei Tos AP. Ewing sarcoma and Ewing-like tumors. Vol. 476, Virchows Archiv. 2020.
4. Gurney JG, Davis S, Severson RK, Fang JY, Ross JA, Robison LL. Trends in cancer incidence among children in the U.S. Cancer. 1996; 78(3).
5. Esiashvili N, Goodman M, Marcus RB. Changes in incidence and survival of ewing sarcoma patients over the past 3 decades: Surveillance epidemiology and end results data. J Pediatr Hematol Oncol. 2008; 30(6).
6. Kano T, Sasaki A, Tomizawa S, Shibasaki T, Tamura M, Ohye C. Primary Ewing's sarcoma of the orbit: Case report. Brain Tumor Pathol. 2009; 26(2).
7. Lai TYY, Leung ATS, Lam DSC, Choi RY, Lucarelli MJ. Primary orbital Ewing sarcoma in a middle-aged woman (multiple letters). Vol. 118, Archives of Ophthalmology. 2000.
8. Agarwal R, Khare A, Gupta N. Isolated Unilateral Orbital Metastasis from Ewing Sarcoma Detected on FDG PET/CT Scan. Clin Nucl Med. 2021; 46(7).
9. Alio JL, Sales-Sanz M, Vaz MA, Barrancos C, Reguero ME, Diamantopoulus J, et al. Primary extraosseous ewing sarcoma of the orbit. Ophthalmic Plast Reconstr Surg. 2013; 29(3).
10. Guzowski M, Tumuluri K, Walker DM, Maloof A. Primary orbital ewing sarcoma in a middle-aged man. Ophthalmic Plast Reconstr Surg. 2005; 21(6).
11. Kaliki S, Rathi SG, Palkonda VAR. Primary orbital Ewing sarcoma family of tumors: A study of 12 cases. Eye (Basingstoke). 2018; 32(3).
12. Widhe B, Widhe T. Initial symptoms and clinical features in osteosarcoma and Ewing sarcoma. Journal of Bone and Joint Surgery. 2000; 82(5).

13. Bacci G, Ferrari S, Rosito P, Avella M, Barbieri E, Picci P, et al. SARCOMA DI EWING DELL'OSSO. STUDIO ANATOMOCLINICO DI 424 CASI. Minerva Pediatr. 1992; 44(7-8).

14. Zöllner SK, Amatruda JF, Bauer S, Collaud S, de Álava E, Dubois SG, et al. Ewing sarcoma—diagnosis, treatment, clinical challenges and future perspectives. Vol. 10, Journal of Clinical Medicine. 2021.

15. Cantile M, Marra L, Franco R, Ascierto P, Liguori G, De Chiara A, et al. Molecular detection and targeting of EWSR1 fusion transcripts in soft tissue tumors. Vol. 30, Medical Oncology. 2013.

16. Tu J, Huo Z, Gingold J, Zhao R, Shen J, Lee DF. The histogenesis of Ewing Sarcoma. Cancer Rep Rev. 2017; 1(2).

17. Setty BA, Gikandi A, DuBois SG. Ewing Sarcoma Drug Therapy: Current Standard of Care and Emerging Agents. Vol. 25, Pediatric Drugs. 2023.

18. Bacci G, Picci P, Gitelis S, Borghi A, Campanacci M. The treatment of localized Ewing's sarcoma: The experience at the istituto ortopedico rizzoli in 163 cases treated with and without adjuvant chemotherapy. Cancer. 1982; 49(8).

19. Klufas MA, Wolden SL, Bohle GC, Wexler LH, Abramson DH. Exenteration and Custom Implant Brachytherapy as a Treatment for Recurrent Primary Extraskeletal Orbital Ewing Sarcoma. Ophthalmic Plast Reconstr Surg. 2015; 31(4).

20. Koontz BF, Clough RW, Halperin EC. Palliative radiation therapy for metastatic Ewing sarcoma. Vol. 106, Cancer. 2006.

21. Abboud A, Masrouha K, Saliba M, Haidar R, Saab R, Khoury N, et al. Extraskeletal Ewing sarcoma: Diagnosis, management and prognosis (Review). Vol. 21, Oncology Letters. 2021.

22. Leavey PJ, Mascarenhas L, Marina N, Chen Z, Krailo M, Miser J, et al. Prognostic factors for patients with Ewing sarcoma (EWS) at first recurrence following multi-modality therapy: A report from the children's oncology group. Pediatr Blood Cancer. 2008; 51(3).

23. Stahl M, Ranft A, Paulussen M, Bölling T, Vieth V, Bielack S, et al. Risk of recurrence and survival after relapse in patients with Ewing sarcoma. Pediatr Blood Cancer. 2011; 57(4).

24. Khan SN, Sepahdari AR. Orbital masses: CT and MRI of common vascular lesions, benign tumors, and malignancies. Saudi Journal of Ophthalmology. 2012; 26(4).