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Embryonic Rhabdomyosarcoma of the Lacrimal Sac: A Rare Presentation and Multimodal Management

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Abstract **Case Report**

Rhabdomyosarcoma (RMS) of the lacrimal sac is an extremely rare and aggressive malignancy arising from mesenchymal cells with skeletal muscle differentiation. While RMS is the most frequent soft tissue sarcoma in children, its manifestation in the lacrimal sac is exceptional and may mimic benign inflammatory conditions, leading to delayed diagnosis and treatment. We report the case of a 45-year-old woman with a rapidly enlarging medial canthal mass, found to be embryonal RMS with orbital and intracranial extension. Diagnosis was confirmed by histopathology and immunohistochemistry. She underwent extensive surgery followed by VAC-IE chemotherapy with good tolerance. Although exceedingly rare, rhabdomyosarcoma of the lacrimal sac should be considered in the differential diagnosis of rapidly progressive orbital masses. This case highlights the rarity and aggressiveness of adult-onset rhabdomyosarcoma of the lacrimal sac, its challenging differential diagnosis, and the importance of a multidisciplinary approach in both diagnosis and management.

Keywords: Rhabdomyosarcoma (RMS); Lacrimal sac; Orbital tumors; soft tissue sarcoma.

INTRODUCTION

Rhabdomyosarcoma (RMS) of the lacrimal sac is an exceptionally rare and aggressive malignant tumor arising from mesenchymal cells with skeletal muscle differentiation [1]. Although RMS is the most common soft tissue sarcoma in children and adolescents [2], its occurrence in the lacrimal sac represents a small fraction of orbital or periorbital tumors [3]. Clinically, it may present with symptoms mimicking benign conditions such as chronic dacryocystitis or a lacrimal sac mucocele [4], often leading to delayed diagnosis. Early identification and a multidisciplinary treatment approach are essential to improve prognosis and preserve ocular function [5–10].

CASE REPORT

We report the case of a 45-year-old female patient with a history of breast neoplasia treated in 2019 with surgery, chemotherapy, and radiotherapy. Three months prior to her follow-up consultation in 2024, she developed a rapidly enlarging mass in the left medial can thus, associated with visual impairment (blurred vision) and ipsilateral nasal obstruction. Clinical examination revealed a firm mass exerting a mass effect on the globe (Figure 1).

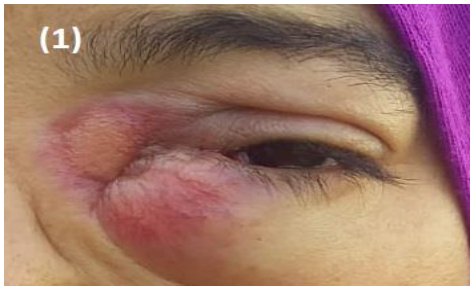

Figure 1: Preoperative photograph: right orbito-palpebral inflammatory swelling, erythematous and indurated

Radiological investigations, including facial CT scan and MRI, revealed a tumour-like lesion involving the lacrimal sac, with endoluminal extension into the ipsilateral intraorbital and endocranial regions, as well as ipsilateral cervical lymph node involvement (Figures 2 and 3).

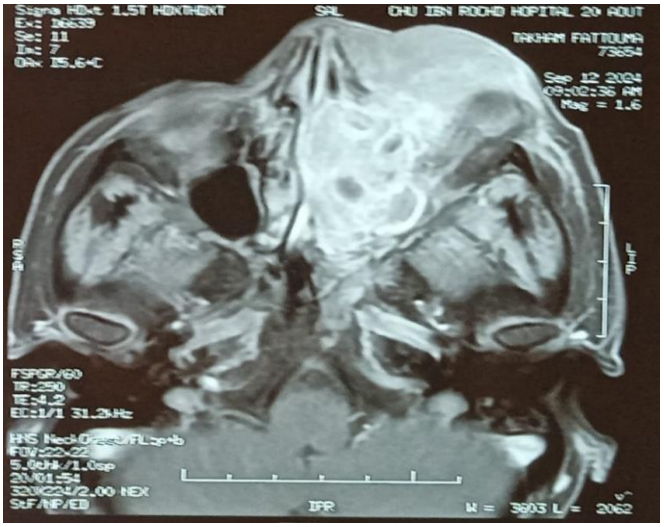

Figure 2: Axial T2-weighted MRI showing a heterogeneous mass in the right lacrimal region with inferomedial displacement of the globe

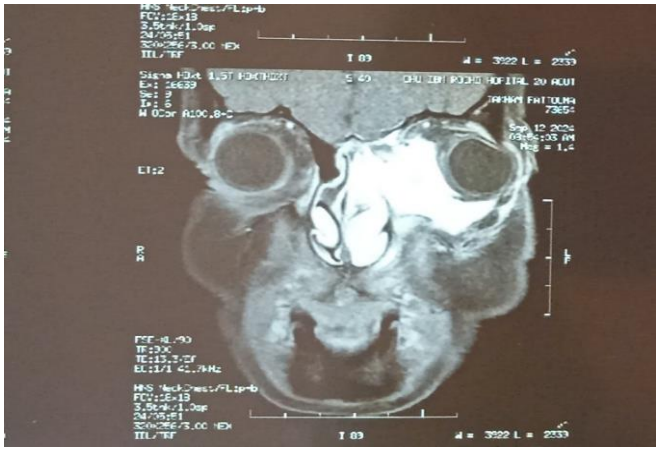

Figure 3: Coronal T2-weighted MRI showing a large, heterogeneous mass in the right lacrimal region extending into the superior orbit, causing inferolateral displacement of the globe

Histopathological analysis of a biopsy specimen revealed a malignant tumour proliferation. Immunohistochemical staining was focally positive for Desmin and Myogenin, and negative for Cytokeratin, Chromogranin, Synaptophysin, and NSE, supporting the diagnosis of an embryonal-type rhabdomyosarcoma of the lacrimal sac (Figure 4).

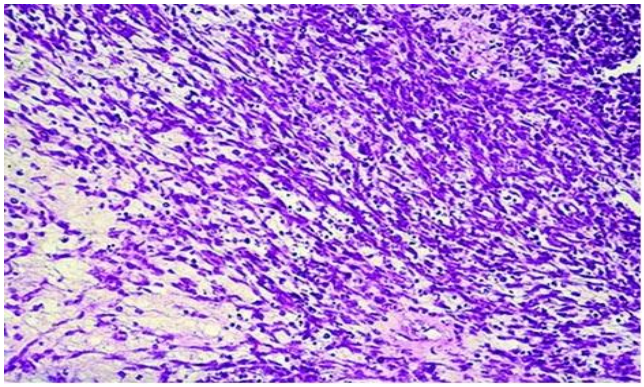

Figure 4: Histological examination. H&E staining (×200): proliferation of round cells with eosinophilic cytoplasm and hyperchromatic nuclei, arranged in sheets and bundles, consistent with embryonal rhabdomyosarcoma

The patient underwent extensive surgical intervention, including tumour resection extending to the maxillary sinus and nasal fossa, left orbital exenteration, parotidectomy, and ipsilateral cervical lymph node dissection (levels II, III, and IV) (Figure 5).

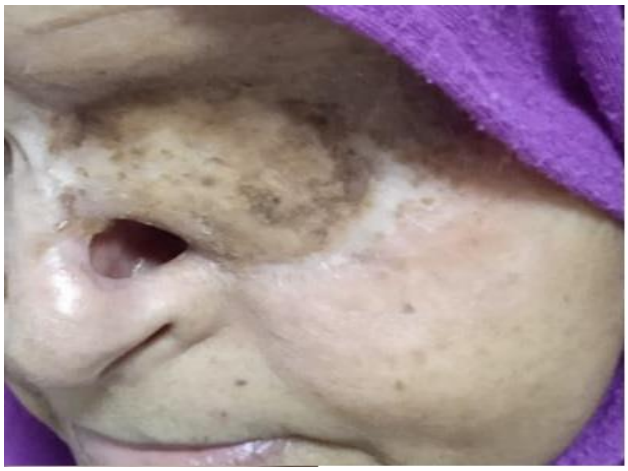

Figure 5: Scar appearance after extended left orbital exenteration, with no visible recurrence

The case was discussed in a multidisciplinary oncology and head-and-neck tumour board (RCP), and the therapeutic decision was to initiate VAC-IE chemotherapy (Vincristine, Actinomycin D, Cyclophosphamide alternating with Ifosfamide and Etoposide). The patient demonstrated good overall tolerance to the treatment.

DISCUSSION

Lacrimal rhabdomyosarcoma is an exceedingly rare malignant tumor originating from the skeletal muscle cells of the lacrimal gland or surrounding orbital tissues. Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children, but its occurrence in the lacrimal gland is exceptional, with only a handful of cases reported in medical literature [1]. This discussion explores the clinical presentation, diagnosis, treatment, and prognosis of lacrimal rhabdomyosarcoma, emphasizing its unique challenges due to its anatomical location and aggressive nature [3].

The clinical presentation lacrimal rhabdomyosarcoma typically presents with rapidly progressive symptoms due to its location in the orbit. Common clinical features include proptosis, periorbital swelling, vision impairment, pain and erythema occur with tumor growth or inflammation [11]. These symptoms often mimic other orbital pathologies, such as orbital cellulitis, lymphoma, or benign lacrimal gland tumors, necessitating a high index of suspicion for accurate diagnosis [12]. The condition predominantly affects children and adolescents, aligning with the demographics of RMS, though adult cases are not unheard of [6].

Diagnosing lacrimal rhabdomyosarcoma requires a multimodal approach combining imaging, histopathology, and staging. Magnetic resonance imaging (MRI) is preferred over computed tomography (CT) for soft tissue characterization, typically showing a heterogeneous mass with possible bone erosion or intracranial extension [13]. Definitive diagnosis relies on a biopsy, which identifies rhabdomyoblasts small, round blue cells with hyperchromatic nuclei and is supported by immunohistochemistry for markers such as desmin, myogenin, and MyoD1 [14,15]. Staging follows the Intergroup Rhabdomyosarcoma Study Group (IRSG) classification to guide therapeutic decisions [16].

Management is based on a multimodal treatment strategy tailored to disease stage and patient factors. Systemic chemotherapy with the standard VAC regimen (vincristine, actinomycin-D, cyclophosphamide) forms the cornerstone of therapy, shrinking the tumor and addressing micrometastases [17]. Radiation therapy provides local control when complete surgical excision is not feasible but carries risks such as cataracts, retinopathy, and facial asymmetry, particularly in children [18]. Surgery is often limited to biopsy or debulking due to the proximity of critical orbital structures, while orbital exenteration is reserved for extensive, unresectable disease [19,20]. Treatment protocols from collaborative groups like the Children's Oncology Group (COG) and the European Pediatric Soft Tissue Sarcoma Study Group (EpSSG) emphasize risk-adapted therapy to balance efficacy and toxicity [21].

The prognosis depends on several factors, including histologic subtype, tumor stage, location, and patient age [22]. Metastatic disease, present in approximately 15–20% of cases at diagnosis, significantly worsens outcomes [23]. Regular follow-up is essential to monitor for recurrence, secondary malignancies, or treatment-related complications [24]. Given the tumor's rarity and the delicate orbital anatomy, lacrimal rhabdomyosarcoma poses unique therapeutic challenges, requiring a balance between tumor control and preservation of vision and cosmesis [25]. Advances

in proton beam therapy may reduce radiation-related morbidity, and targeted therapies—such as those inhibiting the IGF-1R pathway or PAX-FOXO1 fusion proteins—hold promise for improving survival in high-risk patients [26]. International collaboration and registry-based studies remain crucial for refining treatment strategies and optimizing patient outcomes.

CONCLUSION

Rhabdomyosarcoma (RMS) of the lacrimal sac is an extremely rare and aggressive tumor, often presenting with non-specific symptoms such as swelling or discomfort around the medial canthal region. Early diagnosis and prompt intervention are critical in improving patient outcomes. This case underscores the importance of considering RMS in the differential diagnosis of orbital tumors, especially in pediatric and young adult populations. Multidisciplinary management, including surgery, chemotherapy, and radiotherapy, is essential for optimal treatment and improved prognosis. Given the aggressive nature of this malignancy, long-term follow-up is necessary to monitor for recurrence and potential metastasis.

REFERENCES

1. Parham DM, Ellison DA. Rhabdomyosarcomas in adults and children: an update. *Arch Pathol Lab Med*. 2006;130(10):1454-1465. doi: 10.5858/2006-130-1454-RIAACA.
2. Malempati S, Hawkins DS. Rhabdomyosarcoma: review of the Children's Oncology Group (COG) Soft-Tissue Sarcoma Committee experience and rationale for current COG studies. *Pediatr Blood Cancer*. 2012;59(1):5-10. doi: 10.1002/pbc.24118.
3. Shields JA, Shields CL. Rhabdomyosarcoma: review for the ophthalmologist. *Surv Ophthalmol*. 2003;48(1):39-57. doi: 10.1016/S0039-6257(02)00415-0.
4. Burkat CN, Lucarelli MJ. Rhabdomyosarcoma masquerading as acute dacryocystitis. *Ophthalmic Plast Reconstr Surg*. 2005;21(6):456-458. doi: 10.1097/01.iop.0000181349.79287.12.
5. Coffin CM, Dehner LP. Pediatric soft tissue tumors: a clinical, histologic, and immunohistochemical analysis of 200 cases. *Am J Surg Pathol*. 1997;21(5):554-563. doi: 10.1097/00000478-199705000-00002.
6. Sultan I, Qaddoumi I, Yaser S, Rodriguez-Galindo C, Ferrari A. Comparing adult and pediatric rhabdomyosarcoma in the SEER program. *Cancer*. 2009;116(21):5756-5762. doi: 10.1200/JCO.2008.19.7483.
7. Ferrari A, Miceli R, Casanova M, *et al.,* Soft tissue sarcomas in adolescents and young adults: a comparison with their pediatric and adult counterparts. *Lancet Oncol*. 2017;18(3):e166-e175. doi: 10.1016/S1470-2045(17)30099-2.
8. Oberlin O, Rey A, Anderson J, *et al.,* Treatment of orbital rhabdomyosarcoma: survival and late effects of treatment—results of an international workshop. *J Clin Oncol*. 2001;19(1):197-204. doi: 10.1200/JCO.2001.19.1.197.
9. Weiss AR, Harrison DJ. Soft tissue sarcomas in adolescents and young adults. *J Clin Oncol*. 2024;42(6):675-685. doi: 10.1200/JCO.23.01275.
10. Pappo AS. Rhabdomyosarcoma and other soft tissue sarcomas in children. *Curr Opin Oncol*. 1996;8(4):311-316. doi: 10.1097/00001622-199607000-00008.
11. Mouriaux F, *et al.,* Orbital tumors in adults: a retrospective study of 130 cases. *J Fr Ophtalmol*. 2006;29(8):841-846. doi: 10.1016/S0181-5512(06)76545-1.
12. Saboo SS, *et al.,* Imaging features of primary and secondary adult rhabdomyosarcoma. *AJR Am J Roentgenol*. 2012;199(3):W394-W401. doi: 10.2214/AJR.11.7925.
13. Cessna MH, *et al.,* Are myogenin and MyoD1 expression specific for rhabdomyosarcoma? A study of 150 cases. *Am J Surg Pathol*. 2001;25(9):1150-1157. doi: 10.1097/00000478-200109000-00004.
14. Kumar S, *et al.,* Myogenin is a specific marker for rhabdomyosarcoma: an immunohistochemical study. *Mod Pathol*. 2000;13(9):988-993. doi: 10.1038/modpathol.3880174.
15. Lawrence W Jr, *et al.,* Pretreatment TNM staging of childhood rhabdomyosarcoma: a report of the Intergroup Rhabdomyosarcoma Study Group. *Cancer*. 1997;80(6):1165-1170. doi: 10.1002/(SICI)1097-0142(19970915)80:6<1165::AID-CNCR1>3.0.CO;2-3.
16. Crist WM, *et al.,* Intergroup rhabdomyosarcoma study IV: results for patients with nonmetastatic disease. *J Clin Oncol*. 2001;19(12):3091-3102. doi: 10.1200/JCO.2001.19.12.3091.
17. Paulino AC, Okcu MF. Rhabdomyosarcoma. *Curr Probl Cancer*. 2008;32(1):7-34. doi: 10.1016/j.currproblcancer.2007.12.001.
18. Cecchetto G, *et al.,* Biopsy or debulking surgery as initial surgery for locally advanced rhabdomyosarcomas in children? *Cancer*. 2007;110(3):538-544. doi: 10.1002/cncr.22844.
19. Hays DM, *et al.,* Primary reexcision for patients with 'microscopic residual' tumor following initial excision of sarcomas. *J Pediatr Surg*. 1989;24(1):5-10. doi: 10.1016/S0022-3468(89)80242-0.
20. Meza JL, *et al.,* Analysis of prognostic factors in patients with nonmetastatic rhabdomyosarcoma. *J Clin Oncol*. 2006;24(24):3844-3851. doi: 10.1200/JCO.2005.05.3809.
21. Dagher R, Helman L. Rhabdomyosarcoma: an overview. *Oncologist*. 1999;4(1):34-44. doi: 10.1634/theoncologist.4-1-34.
22. Pappo AS, *et al.,* Survival after relapse in children and adolescents with rhabdomyosarcoma. *Cancer*. 1999;86(3):557-564. doi: 10.1002/(SICI)1097-0142(19990801)86:3<557::AID-CNCR19>3.0.CO;2-1.

23. Rodeberg DA, *et al.,* Prognostic factors in childhood rhabdomyosarcoma. *Cancer*. 2001;92(6):1580-1586. doi: 10.1002/1097-0142(20010915)92:6<1580::AID-CNCR1457>3.0.CO;2-3.
24. Donaldson SS, *et al.,* Function preservation and the role of radiation in pediatric orbital rhabdomyosarcoma. *Int J Radiat Oncol Biol Phys*. 1990;18(1):149-156. doi: 10.1016/0360-3016(90)90438-5.
25. Gryder BE, *et al.,* PAX3–FOXO1 Establishes Myogenic Super Enhancers and Confers BET Bromodomain Vulnerability. *Cancer Discov*. 2017;7(8):884-899. doi: 10.1158/2159-8290.CD-16-1297.
26. Koscielniak E, *et al.,* Treatment of soft tissue sarcoma in childhood and adolescence: a report of the German CWS Study Group. *Cancer*. 1999;85(2):396-408. doi: 10.1002/(SICI)1097-0142(19990115)85:2<396::AID-CNCR20>3.0.CO;2-1.