Case Report

Orbital Rosai–Dorfman Disease: A Case Report and Literature Review

Basim Al-Maghrabi, Tarek Elnaggar[1], Osama Alamri[2], Jaudah Al-Maghrabi[3,4]

Departments of Emergency Medicine and [4]Pathology, Faculty of Medicine, King Abdulaziz University, [1]Department of Lid, Lacrimal, Orbit and Oncology, Maghrabi Eye and Ear Hospital, [3]Department of Pathology, King Faisal Specialist Hospital and Research Centre, Jeddah, [2]Department of Ophthalmology, King Abdullah Medical City, Makkah, Saudi Arabia

Abstract

A 53-year-old male presented with dropping of the right eyelid associated with decreased visual acuity for 4 months. He also complained of vertical diplopia especially when looking down. Ophthalmological examination revealed right blepharospasm associated with right hypertropia. There was palpable mass at the inferomedial aspect of the right eye. Magnetic resonance imaging revealed abnormal signal intensity in the right orbit inferior aspect occupying the orbital floor and measured 2.7 cm × 2.5 cm × 1.2 cm and showed enhancement on the postcontrast study. The patient underwent complete excision of the tumor. Histological examination of the mass revealed histiocytic proliferation with emperipolesis, with positive S100, positive CD68, and negative CD1a staining. These histological and immunohistochemical features are consistent with extranodal Rosai–Dorfman disease. There was no complication or recurrence after the complete excision.

Keywords: Emperipolesis, orbital, Rosai–Dorfman disease, sinus histiocytosis with massive lymphadenopathy

Introduction

Rosai–Dorfman disease (RDD) sinus histiocytosis with massive lymphadenopathy is a rare, idiopathic nonhereditary, benign, non-Langerhans cell histiocytosis (LCH), affecting children and young adults, that commonly presents as painless bilateral massive cervical lymphadenopathy with systemic symptoms such as fever, weight loss, and malaise.[1-3] Black people from America, the Caribbean, and West Africa are more frequently affected (43.6%).[4] Extranodal manifestations have been reported in 28%–43% cases.[5] Head and neck involvement represents approximately 22% of extranodal disease.[6] Ocular involvement occurs in 11.5% of RDD cases, often involving orbital soft tissues and the eyelids.[2] Orbit and ocular glove involvement have been reported, usually as a retro-orbital mass and proptosis.[6,7] We report a case of extranodal RDD in a 53-year-old man who presented with right orbital mass.

Case Report

We report a 53-year-old man presented with dropping of the right eyelid associated with a diminished vision for 4 months. He also complained of vertical diplopia especially when looking down. The past medical history revealed chronic sinusitis. There was no previous history of diabetes or hypertension. Ophthalmological examination revealed right blepharospasm associated with right hypertropia. There is palpable mass at the inferiomedial aspect of the right eye. Conjunctiva, cornea, anterior chamber, pupil, iris, and lens were normal. Magnetic resonance imaging revealed orbital mass in the form of abnormal signal intensity in the right orbit inferior aspect occupying the orbital floor, measured 2.7 cm × 2.5 cm × 1.2 cm and showed enhancement on postcontrast study. It was slightly of low-signal intensity on T2, intermediate on T1 and showed enhancement on postcontrast study. Laboratory investigation revealed normal complete blood count.

The patient underwent complete excision of the tumor. The tumor measured 4.5 cm × 3 cm × 0.6 cm. Serial sectioning

Address for correspondence: Dr. Jaudah Al-Maghrabi, Department of Pathology, Faculty of Medicine, King Abdulaziz University, PO Box 80205, Jeddah 21589, Saudi Arabia.
E-mail: jalmaghrabi@hotmail.com

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of the specimen revealed a homogeneous, white tan cut surface. No areas of hemorrhage or necrosis were seen. Microscopic examination revealed aggregates of histiocytes with abundant cytoplasm admixed with numerous plasma cells and some lymphocytes [Figure 1a and b]. Some of the histiocytes appeared to engulf small lymphocytes and plasma cells (emperipolesis). There were fibroblastic proliferation and fibrous tissue formation. The proliferation surrounded and involved nerve bundles, skeletal muscle, and adipose tissue. Immunohistochemistry study revealed that the histiocytes were positive for CD68 and S100 [Figure 1c] and negative for CD1a and langerin. The plasma cells expressed CD79a, CD138 [Figure 1d] and appeared polyclonal with Kappa and Lambda light chain stains. The immunoglobulin G (IgG) 4/IgG ratio was <5% [Figure 1e and f], which against the diagnosis of IgG4-related disease. The lymphoid component was a mixed population of B-cells (CD20 positive) and T-cells (CD3 positive). The fibroblastic proliferation was positive for vimentin and focally for Muscle Specific Actin (MSA). They were negative for desmin, Smooth Muscle Actin (SMA), ALK-1, S100, and pankeratin. Special stains for acid-fast bacilli (Z-N) and fungi (periodic acid–Schiff and Gomori methenamine silver) were negative. These histological and immunohistochemical features are consistent with extranodal RDD. There was no complication after the complete excision. Clinical follow-up until 2 years after surgery did not show any evidence of recurrence.

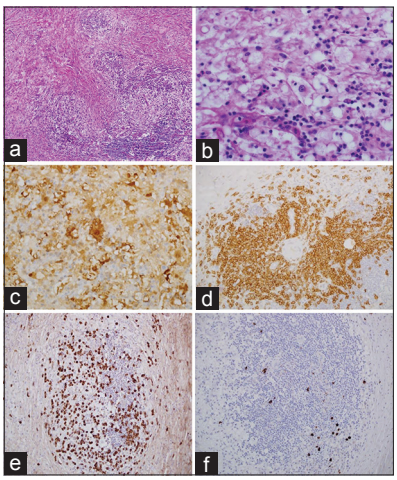

Figure 1: (a) Section from the tumor shows aggregates of lymphohistiocytic proliferation with numerous plasma cells separated by areas of fibrosis (H and E; ×100). (b) Section from the tumor shows aggregates of foamy and vacuolated histiocytes. Some of the histiocytes engulf lymphocytes (emperipolesis) (H and E; ×200). (c) Section from the tumor shows positive nuclear and cytoplasmic S100 immunostaining in the histiocytes (×200). (d) Section from the tumor shows positive CD138 immunostaining in the plasma cells surrounding blood vessels (×200). (e) Section from the tumor shows positive immunoglobulin G immunostaining in the plasma cells (×200). (f) Section from the tumor shows positive immunoglobulin G 4 immunostaining in the plasma cells, and the ratio of immunoglobulin G 4/immunoglobulin G is <5% (×200)

Discussion

Pathogenesis

The exact etiology of RDD is not known; however, according to some authors, the pathogenesis may include recruitment of bone marrow monocytes from peripheral blood into lymph node or extranodal sites and their transformation into the characteristic histiocytes of RDD.[5] Some studies suggest that human herpesvirus type 6 and Epstein–Barr virus may play a role.[8,9] However, until now, nothing has been confirmed to be a direct cause of RDD development.

Pathological findings

The presence of emperipolesis is a hallmark of this disease. Emperipolesis means histiocytes with engulfed lymphocytes (most common), plasma cells, and erythrocytes. Usually, there is associated fibrosis with lymphocyte and plasma cell infiltration. In RDD, the histiocytes characteristically are positive for S100 and CD68 and negative for CD1a.[10] Alpha 1 antichymotrypsin is another histiocytic marker that is also can be positive in RDD.[5] Ultrastructurally, there are no Birbeck's granules which classically seen in LCH.[1,4,11,12] Another rare non-Langerhans histiocytic disorder that is included in the differential diagnosis is Erdheim–Chester disease (ECD), which is typically present as multifocal bony osteosclerotic lesions and characterized pathologically by the presence of foamy histiocytes with multinucleate giant cells. The histiocytes of ECD are usually positive for CD163, CD68, and Factor XIIIa and negative for CD1a and S100, which can differentiate it from RDD. In some cases, the presence of severe fibrosis and few histiocytes in lesions, make the diagnosis of extranodal RDD is more difficult than that of nodal RDD.[13,14] The differential diagnosis also sometimes includes lymphoma, malignant histiocytosis, and monocytic leukemia.[6,7] The differential diagnoses may also include granulomatous inflammatory diseases, inflammatory idiopathic pseudotumors.[15] In the presence of numerous plasma cells, as in our case, the pathological differential diagnosis may also include IgG4-related disease. However, this possibility has been excluded by immunohistochemistry, which demonstrated very low IgG4/IgG ratio.

Laboratory findings

Almost 13% of RDD patients have an associated immune disorder, such as autoantibodies against red cells and joint disease.[4,16] Laboratory abnormalities are nonspecific and may include anemia, hypergammaglobulinemia, and increased erythrocyte sedimentation rate and white blood cell count.[3,5]

Orbital involvement in Rosai–Dorfman disease

Ocular involvement is very rare and is seen in 10% of cases.[4,17] These include eyelid and orbital mass and uveal involvement.[17,18] Most patients with ocular involvement present with proptosis, limited eye movement, eyelid edema, epiphora, and decreased visual acuity. Orbital involvement is usually unilateral and rarely can be bilateral.[3] Infiltrations of the conjunctiva/subconjunctiva, lacrimal tract, cornea,

epibulbar and uveal tract, ciliary body as well as of the optic nerve, have been reported.[2,19,20] In this location, RDD could mimic lymphoma and lacrimal gland tumors.

In the uveal tissue involvement, RDD can be misdiagnosed as malignant melanoma. Therefore, a diagnostic incisional biopsy is recommended in such before considering enucleation.[19] Radiological features of orbital involvement have been reported in a few cases and characterized by infiltrative soft-tissue mass with variable contrast enhancement.[3] When RDD present as a diffuse infiltrating orbital mass, the radiological differential diagnosis includes orbital pseudotumor, lymphoma, sarcoidosis, lymphangioma, or hemangioma.[3] The most frequent radiological appearance of intracranial RDD is a dural-based enhancing mass, mimicking a meningioma.[3]

Treatment

The clinical course of extranodal RDD is typically benign. Many patients experience spontaneously regressing courses; however, some will have disease present in a more extensive form requiring treatment.[3] There is no standard protocol for the treatment of RDD; however, excisional biopsy is frequently done for diagnosis and treatment. There are different treatment modalities for RDD that include surgical excision, radiotherapy, and chemotherapy along with systemic steroids and antimetabolite.[5] In the presence of vital organ compression and/or extranodal localization with important clinical signs, surgical treatment might be the best option.[21,22]

Some patients may succumb to immunologic disorder and infection. The reported mortality rate is 7%, with many RDD patients who have a fatal disease having associated immune dysfunction.[3,23] Complete remission frequently occurs over several months or years;[4] however, recurrence after excisional biopsy may occur.[12]

Conclusion

RDD can present as rarely as an orbital mass. RDD has to be considered in the differentials diagnosis of patients presenting with orbital tumor. Incisional or excisional biopsy is the gold standard tool to confirm the diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Arnao V, Riolo M, Savettieri G, Aridon P. Mercaptopurine treatment in an adult man with orbital and intracranial Rosai-Dorfman disease. Case Rep Neurol Med 2016;2016:1030478.
2. Foucar E, Rosai J, Dorfman R. Sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease): Review of the entity. Semin Diagn Pathol 1990;7:19-73.
3. Wang E, Anzai Y, Paulino A, Wong J. Rosai-dorfman disease presenting with isolated bilateral orbital masses: Report of two cases. AJNR Am J Neuroradiol 2001;22:1386-8.
4. Rosai J, Dorfman RF. Sinus histiocytosis with massive lymphadenopathy. A newly recognized benign clinicopathological entity. Arch Pathol 1969;87:63-70.
5. Shukla E, Nicholson A, Agrawal A, Rathod D. Extra nodal Rosai-Dorfman disease (Sinus histiocytosis with massive lymphadenopathy) presenting as asymmetric bilateral optic atrophy: An atypical ocular presentation. Head Neck Pathol 2016;10:414-7.
6. Goodnight JW, Wang MB, Sercarz JA, Fu YS. Extranodal Rosai-Dorfman disease of the head and neck. Laryngoscope 1996;106:253-6.
7. Nemir J, Trninic I, Duric KS, Jakovcevic A, Mrak G, Paladino J. Extranodal right-optic nerve Rosai-Dorfman disease: A rare localization case report. Surg Neurol Int 2016;7:S1158-62.
8. Pinto DC, Vidigal Tde A, Castro Bd, Santos BH, Ousa NJ. Rosai-Dorfman disease in the differential diagnosis of cervical lymphadenopathy. Braz J Otorhinolaryngol 2008;74:632-5.
9. Li HY, Cui HG, Zheng XY, Ren GP, Gu YS. Orbital Rosai-Dorfman disease in a fifty-eight years old woman. Pak J Med Sci 2013;29:1065-7.
10. Eisen RN, Buckley PJ, Rosai J. Immunophenotypic characterization of sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease). Semin Diagn Pathol 1990;7:74-82.
11. Onciu M. Histiocytic proliferations in childhood. Am J Clin Pathol 2004;122 Suppl:S128-36.
12. Tan JJ, Narang S, Purewal B, Langer PD, Blaydon S, Schwarcz RM, *et al.* Extranodal Rosai-Dorfman disease of the orbit: Clinical features of 8 cases. Ophthalmic Plast Reconstr Surg 2016;32:458-61.
13. Xu Q, Fu L, Liu C. Multimodality imaging-based evaluation of Rosai-Dorfman disease in the head and neck: A retrospective observational study. Medicine (Baltimore) 2017;96:e9372.
14. Yajima M, Nakajima K, Hirato J, Chikamatsu K. Extranodal soft tissue Rosai-Dorfman disease of the head and neck and its diagnostic difficulty. Auris Nasus Larynx 2016;43:345-9.
15. Azoulay R, Brisse H, Fréneaux P, Ferey S, Kalifa G, Adamsbaum C. Lacrimal location of sinus histiocytosis (Rosai-Dorfman-destombes disease). AJNR Am J Neuroradiol 2004;25:498-500.
16. Carbone A, Passannante A, Gloghini A, Devaney KO, Rinaldo A, Ferlito A, *et al.* Review of sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease) of head and neck. Ann Otol Rhinol Laryngol 1999;108:1095-104.
17. Foucar E, Rosai J, Dorfman RF. The ophthalmologic manifestations of sinus histiocytosis with massive lymphadenopathy. Am J Ophthalmol 1979;87:354-67.
18. de Oliveira RC, Rigueiro M, Vieira AC, de Freitas D, Sato E. Rosai-Dorfman disease manifesting as an epibulbar ocular tumour. Clin Exp Ophthalmol 2011;39:175-7.
19. Yousef YA, Al-Hussaini M, Nazzal R, Abdeen G, Alnawaiseh I, Alrawashdeh K. Rosai-Dorfman disease masquerading as uveal melanoma: Case report and review of literature. Hematol Oncol Stem Cell Ther 2018. pii: S1658-3876(18)30061-X.
20. Yataco-Vicente JA, Araujo-Castillo RV, López Fuentes MH. Orbital presentation of Rosai-Dorfman disease. Arch Soc Esp Oftalmol 2018. pii: S0365-6691 (18) 30154-0.
21. Sarwal R, Tu E, Mendelblatt FI, Sugar J, Gross SA, Pulido JS, *et al.* Atypical ocular presentations of Rosai-Dorfman disease. Ocul Immunol Inflamm 2008;16:9-15.
22. Symss NP, Cugati G, Vasudevan MC, Ramamurthi R, Pande A. Intracranial Rosai Dorfman disease: Report of three cases and literature review. Asian J Neurosurg 2010;5:19-30.
23. Goyal A, Mittal A. Destombes-Rosai Dorfman disease: A rare case report. J Maxillofac Oral Surg 2011;10:173-5.