

Case Report

Rhabdomyoblastic Differentiation in Rosai Dorfman Disease of the Orbit in a 12-Year-Old Male

E Ezeanosike, OB Ezeanosike¹, SI Akpan², CN Ezisi

Departments of Ophthalmology and Paediatrics, Federal Teaching Hospital, Abakaliki, Ebonyi State, ²Department of Ophthalmology, University of Uyo Teaching Hospital, Uyo, Akwa Ibom State, Nigeria

ABSTRACT

Extranodal Rosai–Dorfman disease involving the orbit has been reported severally with extensive discourse on management strategies. However, rhabdomyoblastic differentiation coexisting with the disease is yet unreported. We present the clinical and histopathological features of a 12-year-old boy presenting with both. A 12-year-old boy was referred to our Ophthalmic Plastic Surgical Service with a 5-year history of bilateral, progressively enlarging eyelid, orbital masses causing proptosis, and severe cosmetic blemish. He had suffered vision loss to the right eye following trauma 2 years prior to the onset of the disease. Computed tomography of the orbits revealed preseptal and intraorbital components of well-defined multi-lobulated masses in both orbits. No significant nodal involvement was noted. He underwent excision biopsy of the tumor in both eyes performed 1 month apart. A histopathologic diagnosis of the Rosai–Dorfman disease with focal areas of rhabdomyoblastic differentiation was made. He was treated postoperatively with a course of oral steroids with close observation for the rhabdomyoblastic differentiation. Rosai–Dorfman disease is an uncommon disease and has been reported to coexist with other pathologies. We highlight focal myoblastic differentiation within the excised tissues which may not necessarily warrant a diagnosis of rhabdomyosarcoma. Long-term follow-up of this patient is required to establish the safety of observation.

KEYWORDS: Orbit, rhabdomyoblastic differentiation, Rosai–Dorfman disease

Date of Acceptance:

19-Sep-2018

INTRODUCTION

Rosai–Dorfman disease (RDD) was first described in 1965 by Destombes and later in 1969 and 1972 as “Sinus Histiocytosis with massive lymphadenopathy” by Rosai and Dorfman who described the disease as a distinct clinical entity.^[1,2] It was initially described as a disease with predominant lymph node affectation. However, several extranodal involvements of various sites of the body have been published as case reports and small case series.^[3-9]

On gross examination, the tumor appears as a nodular, firm, rubbery, multi-lobulated, and yellowish mass with capsular or pericapsular fibrosis. Microscopically, there is histiocytic expansion or diffuse infiltration by histiocytes. The histiocytes versus the lymphoplasmatic infiltrates create the characteristic light and dark band

appearance. Emperipolesis is a pathognomonic feature characterized by the presence of intact (unphagocytosed) lymphoid cells within the intact histiocytes.^[10]


The disease may occur in the lymph nodes in 57% of cases with 87% of these being cervical lymphadenopathy. Extranodal disease may involve the skin, nasal cavity and paranasal sinuses, soft tissue, orbit, bone, salivary gland, central nervous system, and bone.^[10] Constitutional symptoms and hematological derangement are more common in nodal disease, while extranodal disease presents with symptoms related to the mass effect at the affected location such as proptosis in

Address for correspondence: Dr. E Ezeanosike, Department of Ophthalmology, Federal Teaching Hospital, Abakaliki, Ebonyi State, Nigeria. E-mail: edakspeaksout@gmail.com

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: reprints@medknow.com

How to cite this article: Ezeanosike E, Ezeanosike OB, Akpan SI, Ezisi CN. Rhabdomyoblastic differentiation in rosai dorfman disease of the orbit in a 12-year-old male. Niger J Clin Pract 2018;21:1670-3.

Access this article online	
Quick Response Code:	Website: www.njcponline.com
	DOI: 10.4103/njcp.njcp_262_17

the orbit or airway obstruction in the respiratory tract.^[10] This rare disease often affects younger ages with no racial predilection.^[11] An older report demonstrated a male preponderance of the disease,^[12] but recent larger studies show no sex preference.^[13] Vemuganti *et al.* report an incidence of 0.09% of histopathologically confirmed RDD in over 7000 ophthalmic pathology specimens collected over a 6-year period, indicating its rarity.^[13] Duke and colleagues reported orbital involvement in two siblings from southern Nigeria, and highlighted the familial tendency of the disease.^[14] To the best of our knowledge, this is the first case of RDD of the orbit presenting in our hospital, hence this report.

CASE REPORT

A 12-year-old boy was referred from the South-South region of Nigeria to the Ophthalmic Plastic Surgical Service of the Department of Ophthalmology, Federal Teaching Hospital, Abakaliki with a 5-year history of a painless, slow-growing mass in both orbits causing proptosis. Both eyes had massive preseptal and orbital components. Examination revealed masses that were firm and rubbery in nature, extending from deep in the orbit to the upper and the lower lids in both eyes [Figure 1]. Visual acuity in the right eye was Nil Perception of Light (NPL) following a penetrating ocular injury 2 years prior to the development of the tumor. There was a corneal scar from a sutured laceration with a hazy view of the intraocular details in the right eye. Unaided visual acuity in the left eye was 6/24 with

medial displacement of the globe caused by extensive involvement of the temporal orbit/lateral rectus muscle with the tumor. There were signs of early inferior exposure keratopathy caused by the proptosis for which lubricants were prescribed, and a suture tarsorrhaphy was placed during the surgery for the right eye. The anterior and posterior segment examinations were otherwise normal. A computed tomography (CT) scan of the orbits showed a well-defined multi-lobulated mass extending from the posterior orbit to the lids [Figure 2]. There were no lytic changes in the bony orbit and the mass appeared to be moulding around the orbital structures with expansion of the bony orbits. Excision biopsy was performed for the right orbit via a sub-brow incision for the superior mass and an inferior trans-conjunctival incision for the inferior mass [Figure 3]. The two distinct upper and lower lid masses were removed separately and in toto. An 8–10 mm strip of redundant skin was excised prior to sub-brow wound closure. The inferior trans-conjunctival incision was closed with a single 6.0 vicryl suture. Post-operative antibiotics and oral steroids were commenced. A similar procedure was repeated for the left orbital mass 4 weeks later. The multi-lobulated nature of the single left orbital mass indicated delivery of the tumor as a single unit and this posed a significant challenge leading to multiple dilatations of the pupil from optic nerve contact or compression during surgery. With relief of the pressure as forced surgical rest periods, pupil constricted again. Finally, for the delivery, the preseptal component had to be tucked into the



Figure 1: Pre-operative appearance

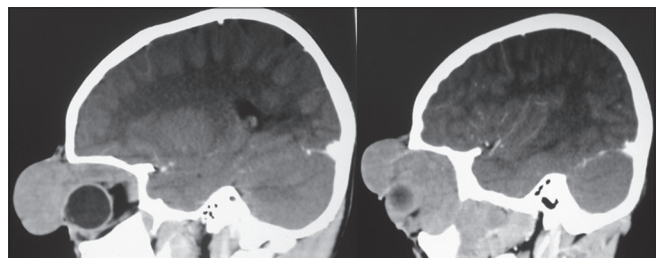


Figure 2: Computed tomography images of the right and left eyes, respectively

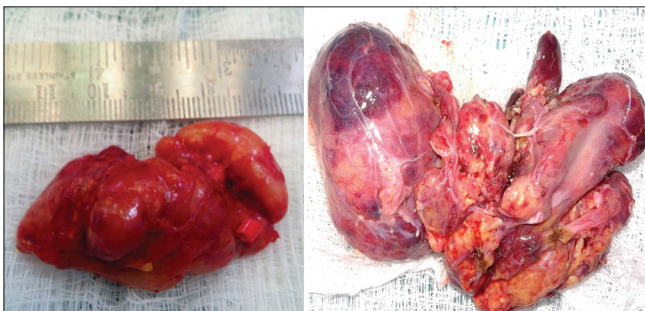


Figure 3: Tumor excised from the right eye and left eye, respectively



Figure 4: Final post-operative appearance

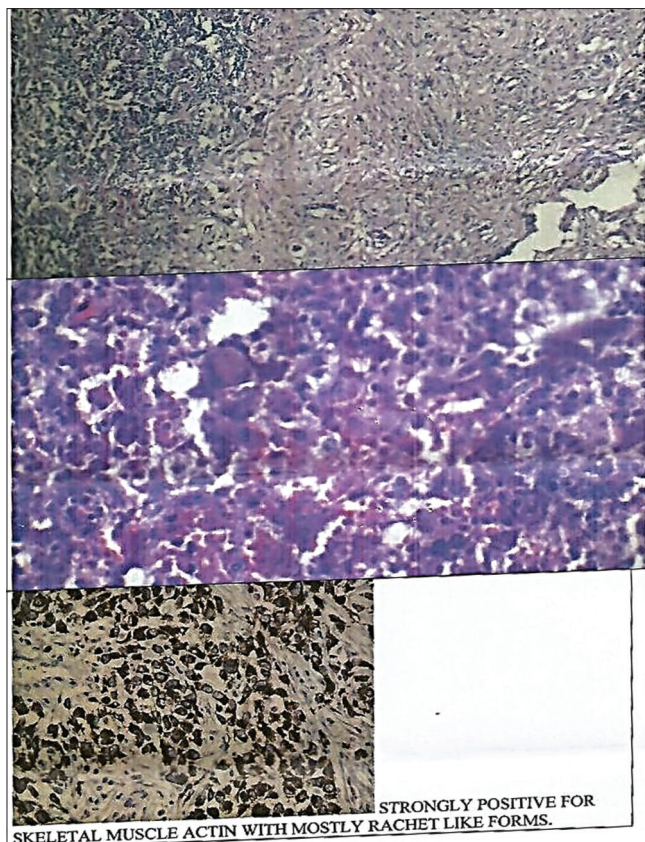


Figure 5: Micrographs for histopathology and immunohistochemistry

orbit and the mass was delivered as a single unit from the inferior trans-conjunctival incision [Figure 3] and closed with a single 6.0 vicryl suture. The skin incision closed in three layers using 6.0 vicryl for periosteum and subcutaneous tissue, while the skin was closed with simple continuous 6.0 Prolene. Adequate wound healing with disappearance of the proptosis was noted following surgery [Figure 4]. Visual acuity on the left eye improved to 6/5 on refraction postoperatively.

Histopathology

The excised tissues from the right eye were two distinct entities – a larger preseptal upper lid mass with orbital extension and a lower lid mass with anterior orbital extension. Histopathology was reported as an atypical pleomorphic rhabdomyoblasts with multinucleated hyperchromatic pleomorphic nuclei, and focal accumulation of lymphocytes and macrophages showing emperipolesis. Immunohistochemical studies were strongly positive for skeletal muscle actin with ratchet-like forms [Figure 5]. An impression of Rosai–Dorfman reaction coexisting with embryonal rhabdomyosarcoma (RMS) was made.

Follow-up

Patient was placed on guttae Maxitriol 2 hourly and tapered over 4 weeks, guttae Ciloxan 4 hourly for

2 weeks, wound cleaning with 5% povidone iodine twice daily, and ointment chloramphenicol over sutures after cleaning for 2 weeks. He received oral ciprofloxacin 500 mg bd for 1 week, ibuprofen 400 mg bd with food for 5 days, and prednisolone at 1 mg/kg/day for 6 weeks and tapered over 4 weeks. No evidence of recurrence has been noted within the 10-month period of follow up.

DISCUSSION

RDD is an uncommon disease. Its etiology remains unknown. The treatment of choice is complete surgical resection with histopathological and immunohistochemical confirmation. Steroid therapy,^[15] chemotherapy, and radiotherapy^[14,16] have also been reported for treatment. Rhabdomyoblastic differentiation in the RDD has not previously been reported. Rhabdomyosarcoma is a malignant tumor of mesenchymal origin,^[17] and is the commonest soft tissue sarcoma of the pediatric age group.^[18] The embryonal type is the commonest (90%) with the best prognosis followed by alveolar, botryoid, or pleomorphic.^[18] The orbital disease presents as a rapidly growing tumor increasing in size over days to weeks and causing proptosis, globe-displacement, a conjunctival, or an eyelid mass.^[17] The duration of this disease in our patient (5 years), its slow progression, and painless bilateral mass lesions are more in keeping with RDD rather than RMS.

A therapeutic dilemma arose for us in the face of the rhabdomyoblastic differentiation as to whether or not to treat. We classified our patient according to the Intergroup Rhabdomyosarcoma Study (IRS) post-surgical staging system as Group 1 (localized disease, completely resected) and Stage 1 (tumor confined to the orbit) which has a good prognosis with treatment with a 5-year survival of up to 94%.^[17] But does the presence of rhabdomyoblasts in a tumor warrant a diagnosis of rhabdomyosarcoma? According to Bishop *et al.*, the presence of rhabdomyoblastic differentiation has been reported in several non-RMS malignancies and does not necessarily imply a diagnosis of RMS.^[19] This was a classic case of Rosai–Dorfman disease (RDD) of the orbit with focal rhabdomyoblastic differentiation. Should all rhabdomyoblastic differentiation be treated? The authors opine that close observation rather than prompt treatment may better serve the interest of the patient. Further intervention would be indicated if there is clinical evidence of disease recurrence.

Ortega *et al.* reports persistent rhabdomyoblastic differentiation on biopsy following completion of therapy for pelvic RMS with no tumor recurrence at a follow up of 3–19 years.^[20] The presence

of rhabdomyoblasts may indicate divergent differentiation and not a metaplastic change and so should be interpreted with caution.^[21] Recurrence following rhabdomyoblastic differentiation in the presence of a background malignant tumor has been reported.^[22] However, Rosai–Dorfman disease (RDD) is a benign tumor with minimal malignant potential. Our consideration was for close surveillance with masterly inactivity in this case. Kroft describes coexistence of RDD with other pathologies with the disease defined by the major component.^[10] Mudhar and Duke report numerous plasma cells, positive for IgG4 in the biopsy sample of their patient with RDD.^[23] Excision biopsy is the mainstay of treatment for orbital cases.^[11]

CONCLUSION

Rosai–Dorfman disease (RDD) is an uncommon disease and has been reported to coexist with other pathologies. Focal myoblastic differentiation within the excised tissues may occur and may not necessarily warrant a diagnosis of rhabdomyosarcoma. Long-term follow-up of this patient is required to establish the safety of observation.

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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

- Rosai J, Dorfman RF. Sinus histiocytosis with massive lymphadenopathy. A newly recognized benign clinicopathological entity. *Arch Pathol* 1969;87:63-70.
- Rosai J, Dorfman RF. Sinus histiocytosis with massive lymphadenopathy: A pseudolymphomatous benign disorder. Analysis of 34 cases. *Cancer* 1972;30:1174-88.
- Campesi G, Daniele E, La Franca S, La Grutta A, Aragona F. The rosai-dorfman lymphadenopathy. Clinical, histological, histochemical and ultrastructural study of a case. *Arch De Vecchi Anat Patol* 1977;62:11-33.
- Foucar E, Rosai J, Dorfman RF. The ophthalmologic manifestations of sinus histiocytosis with massive lymphadenopathy. *Am J Ophthalmol* 1979;87:354-67.
- Walker PD, Rosai J, Dorfman RF. The osseous manifestations of sinus histiocytosis with massive lymphadenopathy. *Am J Clin Pathol* 1981;75:131-9.
- Tanaka T, Asano S, Takahashi K, Miyoshi I, Tanaka M, Hayashi T, *et al.* Sinus histiocytosis with massive lymphadenopathy in a Japanese adult with unusually elevated EBV antibody titers. *Acta Pathol Jpn* 1980;30:121-35.
- Carpenter RJ 3rd, Banks PM, McDonald TJ, Sanderson DR. Sinus histiocytosis with massive lymphadenopathy (Rosai-dorfman disease): Report of a case with respiratory tract involvement. *Laryngoscope* 1978;88:1963-9.
- Sanchez R, Sibley RK, Rosai J, Dorfman RF. The electron microscopic features of sinus histiocytosis with massive lymphadenopathy: A study of 11 cases. *Ultrastruct Pathol* 1981;2:101-19.
- Avril MF, Amiel JL, Théodore C, Chahine G, Caillaud JM, Caillou B, *et al.* Skin manifestations of the rosai-dorfman syndrome. *Ann Dermatol Venereol* 1984;111:661-2.
- Kroft SH. Rosai-dorfman disease: Familiar yet enigmatic. *Semin Diagn Pathol* 2016;33:244-53.
- 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.
- Stones DK, Havenga C. Sinus histiocytosis with massive lymphadenopathy. *Arch Dis Child* 1992;67:521-3.
- Vemuganti GK, Naik MN, Honavar SG. Rosai dorfman disease of the orbit. *J Hematol Oncol* 2008;1:7.
- Duke R, Asana U, Nkanga D, Ibanga A, Mudhar H. Rosai-dorfman disease in African children. *Internet J Ophthalmol Vis Sci* 2009;8:1-5.
- Ocheni S, Ibegbulam OG, Okafor OC, Raveenthiran V, Nzegwu MA. Usefulness of oral corticosteroid in rosai-dorfman disease. *Eur J Cancer Care (Engl)* 2007;16:286-8.
- 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.
- Jurdy L, Merks JH, Pieters BR, Mourits MP, Kloos RJ, Strackee SD, *et al.* Orbital rhabdomyosarcomas: A review. *Saudi J Ophthalmol* 2013;27:167-75.
- Shields CL, Shields JA, Honavar SG, Demirci H. Primary ophthalmic rhabdomyosarcoma in 33 patients. *Trans Am Ophthalmol Soc* 2001;99:133-42.
- Bishop JA, Thompson LD, Cardesa A, Barnes L, Lewis JS Jr., Triantafyllou A, *et al.* Rhabdomyoblastic differentiation in head and neck malignancies other than rhabdomyosarcoma. *Head Neck Pathol* 2015;9:507-18.
- Ortega JA, Rowland J, Monforte H, Malogolowkin M, Triche T. Presence of well-differentiated rhabdomyoblasts at the end of therapy for pelvic rhabdomyosarcoma: Implications for the outcome. *J Pediatr Hematol Oncol* 2000;22:106-11.
- Govender D, Pillay P. Primary myxoid liposarcoma with rhabdomyoblastic differentiation. *Arch Pathol Lab Med* 1998;122:740-2.
- Shanks JH, Banerjee SS, Eyden BP. Focal rhabdomyosarcomatous differentiation in primary liposarcoma. *J Clin Pathol* 1996;49:770-2.
- Mudhar HS, Duke R. A case of orbital rosai-dorfman disease with IgG4 positive plasma cells. *Orbit* 2013;32:315-7.