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Case Report

Extranodal right-optic nerve Rosai–Dorfman disease: A rare localization case report

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Abstract

Background: Rosai–Dorfman is a rare disease that usually occurs in young adults. It is characterized with massive painless cervical lymphadenopathy and histiocyte proliferation. Isolated intracranial involvement is extremely rare. Our aim is to present a new rare case of extranodal Rosai–Dorfman disease that involved the right optic nerve in a 4-year-old boy.

Case Description: A 4-year-old boy with right-sided convergent strabismus and amblyopia lasting for 1 year was treated at the Department of pediatric ophthalmology. Initial optical fundus examination was normal. Examination repeated after 1 year noted the atrophy of the optic nerve papilla. Visual evoked potentials of the right eye showed normal findings of prechiasmatic visual pathway with severe dysfunction of the right optic nerve. Magnetic resonance imaging (MRI) of the brain and orbits showed expansive changed and elongated right optic nerve with contrast enhancement, and smaller lesion in the right temporal operculum region visible in T2 and fluid-attenuated inversion recovery sequence. Through small eyebrow "keyhole" osteoplastic frontoorbital craniotomy the fusiform enlarged (to 2 cm) right optic nerve was identified, resected between the eyeball and optic chiasm, and transferred for pathohistological analysis. Early postoperative course had no complications. Histological, immunohistochemical, and ultrastructural analyses revealed extranodal Rosai–Dorfman disease. Right periorbital edema was verified on the 7th postoperative day and regressed to supportive therapy. Control multi slice computed tomography (MSCT) and MRI of endocranium and orbits showed total tumor removal with no signs of complications.

Conclusion: Although rare, extranodular intracranial Rosai–Dorfman disease should be taken into account in the differential diagnosis of intracranial and intraorbital lesions, especially in the pediatric age group.

Key Words: Extranodal, optic nerve, pediatric tumor, Rosai–Dorfman disease

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INTRODUCTION

Rosai–Dorfman disease (RDD), also known as sinus histiocytosis with massive lymphadenopathy (SHML) is an uncommon benign histiocytic proliferative disorder of unknown origin.[1,2,7,9,12,14,19] It predominantly affects the lymph nodes but can also be found extranodally in other

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organs, and usually presents with other constitutional symptoms such as fever, malaise, weight loss, and raised inflammatory markers.[1,2,7,9,12,14,19] Approximately, one-third of the patients have concurrent extranodal involvement, most commonly in the skin, salivary glands, and upper respiratory tract.[21] Nervous system involvement is rare and in most cases intracranial.[2,9,11,12] Only 18 cases of intracranial involvement have been described previously. In addition, only 4 cases described the lesion with involvement of the orbits. Our report documents the first case, to our knowledge, of optic nerve localization of RDD.

CASE DESCRIPTION

History and examination

A 4-year-old boy presented with right-sided convergent strabismus and amblyopia that lasted for 1 year and was admitted to the Department of Pediatric Ophthalmology. His medical history was unremarkable except for the thrombocytopenia treated at 2 years of age. Before admission, he had regular ophthalmological exams and was treated with corrective glasses. An eye examination was performed by an ophthalmologist. Visual acuity was normal on his left eye, but visual acuity on his right eye was poor, he did not have the sense of light. Both eye bulb motility was normal with good pupil function. Initial optical fundus examination was normal. The cover test was positive on his right eye with right-sided convergent strabismus. Visual field was not done because of the patient's age. On the last ophthalmologic control exam, due to the right-sided atrophy of the optic nerve papilla and amblyopia, performing visual evoked potentials (VEP) and magnetic resonance imaging (MRI) were recommended. VEP of the right eye showed normal findings of prechiasmatic visual pathway with severe dysfunction of the right optic nerve. MRI of the brain and orbits showed expansive changed and elongated right optic nerve with contrast enhancement; furthermore, a smaller lesion in the right temporal operculum region was visible in T2 and fluid-attenuated inversion recovery sequence, as well as a small oval lesion in the left cerebellar lobe [Figures 1 and 2]. Optic nerve glioma was considered to be the most likely radiological diagnosis.

Surgical technique

The position of the patient was supine, with the head turned to the left at 15°, leaving the eyebrow as the most prominent point. Skin was incised through the eyebrow, medially up to the supraorbital notch leaving the supraorbital nerve intact. With one small burr hole at the superior temporal line, small supraorbital bone flap was performed which was 3 cm in width and 2.5 cm in height using a craniotome, including linear extensions over the supraorbital arch. Periorbita was detached from the bone and the bony flap was pushed down toward the orbit until the orbital roof brakes. With slight elevation of the bone, dura was detached from the orbital roof and the whole bone flap including supraorbital arch was removed in one piece. Using a diamond drill, the whole orbital roof was drilled out, optic canal widely opened, and anterior clinoid removed extraduraly. Periorbit was incised longitudinally, including annulus of Zinni and the dura over the optic nerve and frontobasaly, meticulously dissecting orbital muscles and nerves before reaching the optic nerve. Almost the whole optic nerve was thickened, and was irregularly shaped 2 mm from the eyeball and up to the chiasm. The nerve with the infiltrating tumor was cut leaving the normal white tissue at the resected planes of the nerve. Macroscopically, the tumor was firm, avascular, and had a gray-yellow color infiltrating the whole width of the optic nerve. There was no bleeding at the resected planes of the nerve and dura and periorbit was partly sutured and sealed. Bone flap was attached using microscrews, and the wound was closed in the standard manner.

Postoperative course

Early postoperative course was uneventful, except right-sided periorbital hematoma that spontaneously regressed few days later. Later, the patient received corneal ulcer refractory to treatment with topical antibiotic drops and cream for 3 months, after which the amniotic membrane transplant was performed to heal the ulcer completely. After 3 years of follow-up, MRI showed

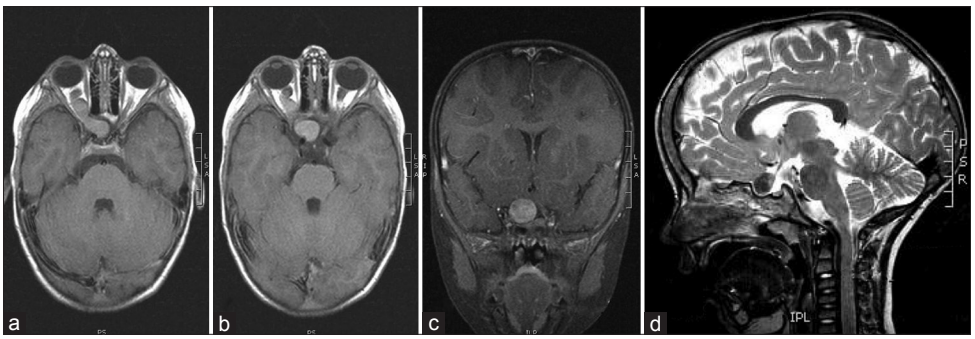

Figure 1: (a, b) Preoperative T1 magnetic resonance (MR) images demonstrates expansive changed and elongated right optic nerve. (c) Preoperative coronal plane T1 gadolinium-enhanced MR image. (d) Preoperative sagittal plane T2 MR image

complete tumor removal without any sign of recurrence [Figure 3].

Histopathological workup

Microscopic examination of the surgical sample revealed a mainly histiocytic lesion in a fibrous background admixed with lesser populations of lymphocytes and plasma cells. The histiocytic cells showed evidence of emperipolesis. Immunohistochemically, tumor cells were CD68 and S100 positive and negative for langerin and CD1a. The phenotype was consistent with RDD [Figures 4 and 5]. There were no microorganisms, necrosis, or granuloma formation. Because of the extremely rare diagnosis, especially on this localization, paraffin blocks of tumor tissue were sent to the Department of Pathology Brigham and Women's Hospital in Boston, USA, for a second opinion. They confirmed our diagnosis of RDD.

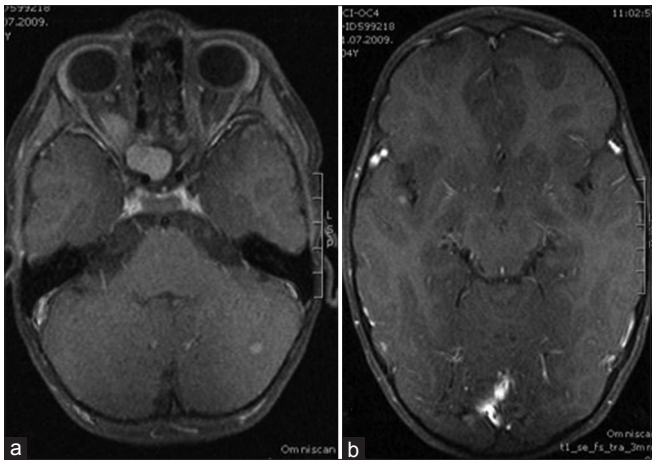

Figure 2: (a, b) Preoperative magnetic resonance imaging showing left cerebellar and right temporal intracranial lesions

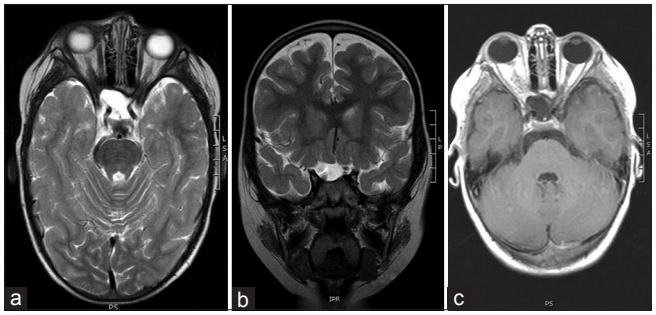

Figure 3: (a,b) Postoperative T2 images, axial and coronal plane. (c) Postoperative T1 axial plane image

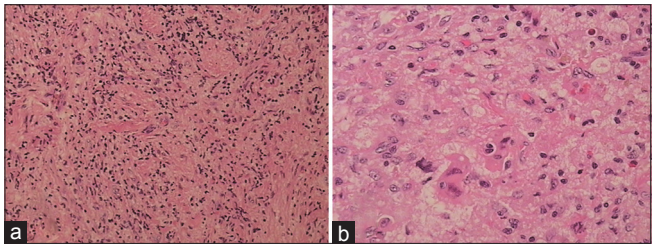

Figure 4: (a) Microscopic features; mainly histiocytes and a few chronic inflammatory cells in fibrous background (hematoxylin and eosin ×100). (b) Emperipolesis; an additional common histopathologic finding in Rosai–Dorfmann disease (hematoxylin and eosin ×400)

DISCUSSION

RDD is a rare histiocytic disorder initially described as a separate entity in 1969 by Rosai and Dorfman using the term SHML.[19] The causes of RDD are not fully understood, and treatment strategies can be different according to the severity of vital organ involvement. It is usually seen in young adult patients but may occur in any age group.[13] Patients presenting with isolated intracranial disease tend to be older.[6] Moreover, it is more common in men, with a possible predilection for African Americans, and the mean age at presentation is approximately 20 years.[7,9,19] The etiology is uncertain, although agents such as the Epstein-Barr or herpes viruses are important in the pathogenesis.[10] The neck lymph nodes are the most frequently involved, followed by inguinal, axillary, and mediastinal lymph nodes.[3,16] The most common extranodal sites are the skin, upper respiratory tract, and bones. Head and neck involvement—approximately 22% of extranodal disease—include involvement of the nasal cavity, the paranasal sinuses, the nasopharynx, submandibular glands, the parotid, the larynx, the temporal bone, the intratemporal fossa, the pterygoid fossa, the meninges, and the orbit.[3,18] The skin is also commonly affected. Half of the patients have another associated extranodal site. Orbit and ocular glove involvement have been reported, usually as a retroorbitary mass and proptosis.[8] To our knowledge, the present case is the first example of optic nerve localization caused by RDD. Intracranial RDD usually occurs without extracranial lymphadenopathy, and most intracranial lesions are attached to the dura with only few extending intraparynchemally. Central nervous system disease can present clinically and radiologically as meningioma, however, the presence of emperipolesis in the cerebrospinal fluid is usually diagnostic of RDD.[15] In our case, glioma was considered to be the most likely radiological diagnosis. Emperipolesis is not a unique phenomenon to RDD and has been seen in both normal and leukemic processes,[19] however, it appears to be a prerequisite for the diagnosis. The clinical course of RDD is unpredictable with episodes of exacerbation and remissions that could last many years. The disease is often self-limiting with a very good outcome, nevertheless 5–11% of patients die from their disease. In the present case, clinical presentation of RDD was right-sided convergent strabismus and amblyopia without painless cervical lymphadenopathy with fever, which has been seen in other patients. The presenting

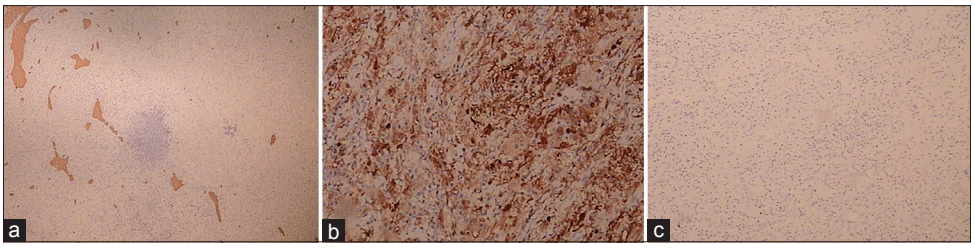

Figure 5: Immunohistochemistry: (a) Glial fibrillary acidic protein small fragments of brain tissue were positive whereas tumour was negative (×100). (b) CD68 positive histiocytic tumor cells (×400); (c) negative staining for CD1a (×100)

symptoms depended on the location of the lesions and were manifested by cranial nerve deficits or nonlocalizing symptoms of raised intracranial pressure or seizures. Laboratory features in RDD are often nonspecific. Leukocytosis, elevated sedimentation rate, and polyclonal hypergammaglobulinemia have been reported in most patients, but in our case there were no laboratory abnormalities.[7,20] The differential diagnosis of extranodal SHLM may be a challenge, and is based on the clinical and histological examination. Histology shows typical features, such as diffuse lymphoplasmatic infiltration, Russel bodies, foamy histiocytes, and histiocytes with phagocytosed lymphocytes within the cytoplasm (emperipolesis). Immunohistochemical features include positive S-100, alpha-antichymotrypsin and CD1a and CD68 antigens.[3,18] Imaging (computed tomography and MRI) may be used to assess disease extension. If there is cervical lymph node enlargement, fine needle aspiration biopsy or lymph node biopsies may be useful for the diagnosis.[10] The differential diagnosis is made with lymphoreticular malignancies such as lymphomas, Hodgkin's disease, malignant histiocytosis, and monocytic leukemia, all of which have similar histopathological features. Atypia in cytology and the aggressive clinical course establish the diagnosis in most case. Other histiocytoses, such as rhinoscleromas and Wegener's granulomatosis, may also be included in the differential diagnosis.[8] Treatment is controversial. In the majority of cases, RDD has a benign course and treatment is not necessary.[22] Therapy is required, however, for patients with extranodal RDD having vital organ involvement or those with nodal disease causing life-threatening complications.[18] The role of surgery is mostly in biopsies and to relieve obstruction.[10] Local recurrence is frequent following surgical resection. The role of radiotherapy is not well understood; some reports have described full resolution with this treatment, whereas others have shown no response.[4,5] Systemic corticosteroids are usually helpful in decreasing nodal size and symptoms, however, they can be immunosuppressive and recurrence of RDD lesions can occur after a short period of interruption.[21] Chemotherapy has resulted in controversial results. A possible efficacy of methotrexate and 6-mercaptopurine requires further investigation. Other reports have suggested using alpha-interferon, although its side effects have its limited use.[14] Furthermore, the efficacy of the anti-CD20 monoclonal antibody/rituximab has been described in one case.[17] We treated our patient with the surgical form of treatment, to which he responded well, without radiotherapy and others modalities. Because of the rarity of such cases, long-term clinical and radiological follow-up is mandatory. In summary, we described a new case of isolated optic nerve RDD causing optic nerve infiltration. It should be considered among the rare differential diagnosis of optic nerve infiltration disease.

CONCLUSION

Extranodular intracranial RDD should be taken into account in the differential diagnosis of intracranial and intraorbital lesions, especially in pediatric age group.

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Conflicts of interest

There are no conflicts of interest.

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