

Refractory extra-nodal Rosai-Dorfman disease of cheek and upper eyelid: A rare case

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Abstract

Rosai-dorfman disease (RDD) is a rare benign disorder of histiocytes characterized histologically by intracellular engulfment of lymphocytes. Extra-nodal RDD has been reported in 43% of cases. However, soft tissue cheek mass is rare presentation of extra-nodal RDD. A 31-year-old female presented with a history of right cheek mass and right upper eyelid mass gradually increasing for 8 months, with no palpable cervical lymphadenopathy. The masses were excised and sent for histopathological examination. The histiocytes showed positive immunostaining for S100 and negativity for CD1a, consistent with extra-nodal RDD. Patient was on systemic steroids and was doing well. After 12 months of follow-up, patient had recurrence in right cheek, which was again excised. Again the mass recurred after 10 months, was excised and patient was put on radiotherapy to the cheek mass. Extra-nodal RDD with involvement of cheek is a rare presentation. Incorporation of S100 and CD1a is helpful in diagnoses of RDD and differentiating it from other benign histiocytosis. The causes of RDD are not fully understood, and treatment strategies can be different according to severity or vital organ involvement.

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1. Introduction

Rosai-Dorfman disease (RDD) is a rare histiocytic disorder initially described as a separate entity by Rosai and Dorfman under the term sinus histiocytosis with massive lymphadenopathy. The etiology of sinus histiocytosis with massive lymphadenopathy (SHML) remains unknown. Although the clinical manifestations and histologic appearance are suggestive of an infectious process, no microorganism has yet been identified. In addition to involving lymph nodes, still the principal site, in approximately one-third of patients, SHML, in the form of pseudotumors, can occur in a variety of extra-nodal sites.^[1] Virtually any extra nodal site can be affected, but the head and neck region (including the sinuses, orbit and ear) is most common.^[2] Other relatively commonly involved extra nodal sites include the soft tissue, skin, upper respiratory tract, gastrointestinal tract, breast, bones, and the central nervous system. However, Rosai-Dorfman disease presenting as recurrent cheek mass without palpable cervical lymph nodes, is rare. Herein we are reporting a 31-year-old female presenting with recurrent extra-nodal RDD of cheek, which is refractory to excision and steroid therapy.

2. Case Report

A 31-year-old Indian female presented with history of right cheek mass and right upper eyelid swelling gradually increasing for 8 months, with no palpable cervical lymphadenopathy. The masses were excised and sent for histopathological examination. The cheek mass measured 7.5 x 6.0 x 5.1 cm (Figure 1) and the right upper eyelid mass measuring 3.0 x 2.7 x 2.0 cm. The masses were serially sliced, the cut surface was homogenous gray-white, representative sections were submitted (Figure 2).



Figure 1: The gross picture of cheek mass measuring 7.5 x 6.0 x 5.1 cm.



Figure 2: The cut surface was homogenous gray-white.

Sections showed mixed population of cells, including lymphocytes, plasma cells, and histiocytes (Figure 3, 4). The most characteristic cells were large and irregular histiocytes, with abundant, acidophilic, sometimes vacuolated cytoplasm, vesicular nucleus and occasional central nucleolus.

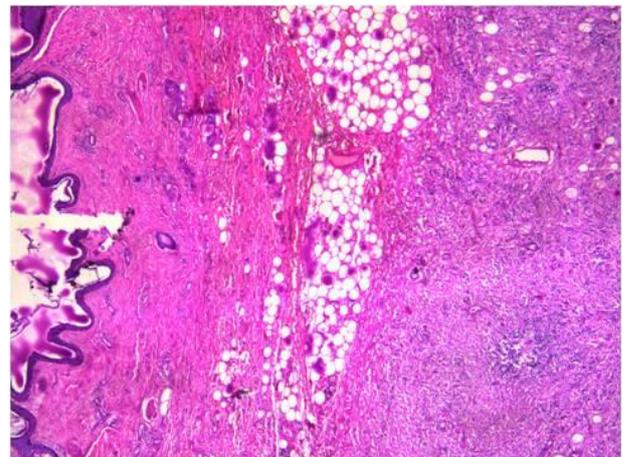


Figure 3: Histologic picture showing a diffuse mass in deep dermis (H&E, x100).

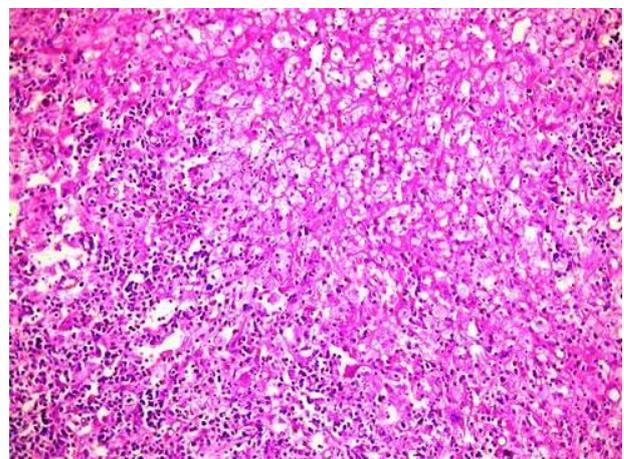


Figure 4: Histologic picture showing sheets of histiocytes admixed with plasma cells & lymphocytes (H&E, x200).

Mitoses were rare. The intracytoplasmic vacuoles contain engulfed cells, usually lymphocytes, plasma cells, or erythrocytes showing 'emperipolesis' (Figure 5). Other cells include numerous plasma cells, mature lymphocytes; and occasionally large, compact aggregates of lipid-laden histiocytes. Eosinophils were rare. Necrosis was not identified. The histiocytes showed positive immunostaining for S100 (Figure 6) and negativity for CD1a, consistent with extra-nodal RDD. Incorporation of S100 and CD1a was helpful in diagnoses of RDD and differentiating it from other benign histiocytosis. As no atypia or mitotic activity was present, malignant histiocytic tumors were excluded. Patient was on systemic steroids and was doing well. After 12 months of follow-up, patient had recurrence in right cheek, which was again excised. Again the mass recurred following 10 months, was excised, facial nerve was compromised and patient was put on radiotherapy to the cheek mass. The repeated recurrence of the cheek mass despite surgery and steroid therapy shows the refractory nature of the mass.

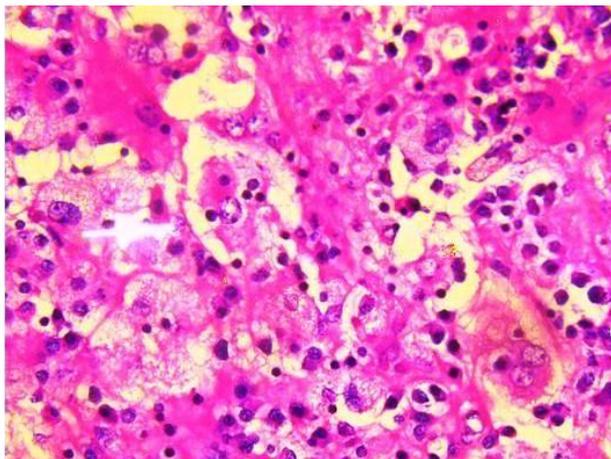


Figure 5: Histologic picture showing emperipolesis (H&E, x400).

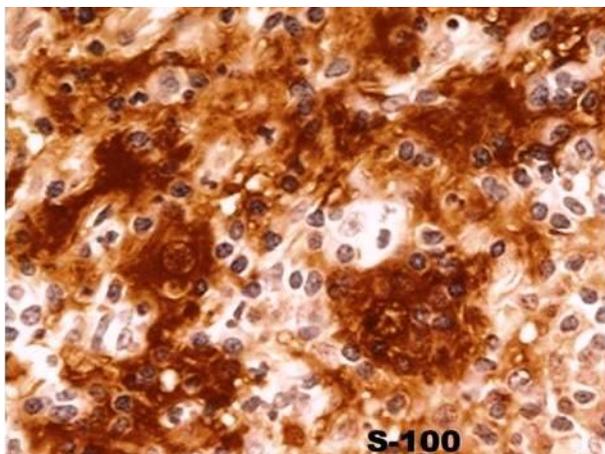


Figure 6: Immunohistochemistry showing S100 positivity. (S-100, DAB, X400).

3. Discussion

RDD, also known as sinus histiocytosis with massive lymphadenopathy, is a rare disease that was first described by Destombes in 1965 and recognized as a distinct clinicopathologic entity by Rosai and Dorfman in 1969.^[3,4] In its classic form, the disease presents in children and adolescents with massive painless cervical lymphadenopathy, often with associated fever, mild anemia, polyclonal hypergammaglobulinemia, and an elevated erythrocyte sedimentation rate.^[1] Microscopically, enlarged lymph node sinuses contain prominent numbers of histiocytes with phagocytosed lymphocytes.^[5] The mean age of onset of nodal disease is 20.6 years, with a male-to-female ratio of 1.4:1. Patients who present with or subsequently develop intracranial involvement, however, become symptomatic at a significantly later mean age (34.9 years), with a strong male predominance.^[6] Its variant extra-nodal RDD is seen in 43% cases of RDD.^[1] The histological findings in extra-nodal RDD are characterized by dense infiltrate of histiocytes with scattered lymphocytes, plasma cells, and neutrophils in the dermis or subcutaneous tissue. The histiocytes are larger in size with large vesicular nuclei, small nucleoli, and abundant pale pink cytoplasm. Emperipolesis (the presence of intact lymphocytes, plasma cells, neutrophils, and red blood cells) within histiocytes is the pathognomic feature.^[7] Immunohistochemistry shows the positivity for S-100 and CD68 and the negativity for CD1a.^[8] Our patient was 31-years-old female, presenting primarily with cheek mass and upper eyelid swelling without lymph node involvement. The histopathological and immunohistochemical features were similar to the above finding.

The exact pathogenesis of the disease is not known. Treatment should be based on severity of symptoms. No ideal treatment guidelines exist for the RDD. Many RDD lesions remain asymptomatic and heal spontaneously without any intervention.^[9] Surgical excision, laser excision, steroids, liquid nitrogen, alkylating agents, thalidomide, isotretinoin and radiation therapy have been used with variable outcomes.^[10] In our patient initially response was achieved with surgical excision followed by steroids. However, patient had repeated recurrence in the same site with those modalities of treatment. So the newer modalities of treatment were considered like Rituximab and Irradiation. Radiation therapy was known to have limited efficacy in most cases of RDD, although recent reports have shown benefit in a refractory extra-nodal RDD with higher radiation doses.^[11] So, after discussions in multidisciplinary team meetings, the patient was referred for Radiotherapy.

4. Conclusion

To conclude, steroid-resistant refractory extra-nodal RDD of cheek is rare manifestation and can be difficult to manage. Multidisciplinary approach is absolutely necessary to achieve disease control. The pathologist must be aware of the unusual presentation of the disease and also the possible differential diagnosis. Early diagnosis by pathologist is very important to obtain proper treatment decision and to improve the quality of life in such patients.

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Competing Interests

None declared.

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