International Journal of Contemporary Pediatrics
Goyal S et al. Int J Contemp Pediatr. 2025 Jul;12(7):1226-1228
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

Case Report

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20251880

Rosai Dorfman disease: a rare entity

Sweety Goyal[1]*, Chandra M. Sharma[1], Cheena Garg[2]

[1]Department of Pediatrics, Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, India
[2]Department of Pathology, Rohilkhand Medical College and Hospital, Bareilly, Uttar Pradesh, India

Received: 22 April 2025
Revised: 14 May 2025
Accepted: 22 May 2025

***Correspondence:**
Dr. Sweety Goyal,
E-mail: sweetysgoyal2014@gmail.com

ABSTRACT

Rosai Dorfman disease (RDD), a rare benign proliferative disorder of unknown etiology, stems from abnormal histiocyte accumulation within lymph node sinusoids and extra nodal tissues. While uncommon in paediatric population, RDD clinically simulates lymphoproliferative disorders. This report details a 4-year male patient presenting at a tertiary care centre with progressive bilateral cervical and submandibular lymphadenopathy lacking febrile symptoms. Diagnostic confirmation was achieved through excisional biopsy. RDD is an exclusion-based diagnosis which requires multidisciplinary collaboration and heightened clinical suspicion remains essential for paediatricians and oncologists to facilitate timely intervention and management of this rare entity.

Keywords: Lymph nodes, Histopathology, Non-Langerhan cells, Steroids

INTRODUCTION

Rosai-Dorfman disease (RDD), classified among non-langerhan cell histiocytosis, represents a diagnostically challenging condition.[1] Alternatively termed "sinus histiocytosis with massive lymphadenopathy". The condition demonstrates male predominance, with a gender ratio approximating 2:1 and global incidence nearing one in 200,000. This benign systemic histioproliferative disorder primarily affects lymphatic architecture. Cervical involvement remains the most well-documented presentation, though peripheral and central lymph node engagement may occur.[2] Additional manifestations include mediastinal, axillary, inguinal and retroperitoneal lymphadenopathy. Extra nodal involvement emerges in twenty-five to forty percent of cases, frequently involving cutaneous, respiratory, genitourinary, skeletal, orbital, central nervous system (CNS) and mammary systems.[3] CNS manifestations affect fewer than five percent of patients, with seventy-five percent presenting as intracranial lesions versus twenty-five percent as spinal involvement. Pathognomonic histopathological features include CD68(+), S100(+), CD1a(-) histiocyte aggregates demonstrating emperipolesis.[1]

CASE REPORT

A 4-year male patient presented at the paediatric OPD with multiple neck swellings persisting for 1 year (Figure 1). These non-tender masses demonstrated progressive enlargement without associated fever, night sweats or weight loss. Clinical evaluation revealed stable vitals alongside multiple firms, non-matted cervical lymph nodes: pre-auricular (3 cm), post-auricular (2.8 cm), jugular (3 cm) and infra-auricular (1 cm and 0.5 cm). Inguinal and axillary regions remained unremarkable. Laboratory analysis indicated haemoglobin levels of 9.2 gm/dl, total leukocyte count (TLC) 8000 and erythrocyte sedimentation rate (ESR) 63 mm. Thoracic imaging demonstrated hilar lymphadenopathy on plain radiography (Figure 2), corroborated by HRCT findings of multifocal nodal enlargement across mediastinal stations, largest measuring 15 mm in SAD (Figure 3). Abdominal ultrasound showed no pathological features. Histopathological examination following excisional

biopsy demonstrated dense lymphohistiocytic proliferation with characteristic sinus histiocytes exhibiting emperipolesis (Figure 4). Absence of langerhan cells, granulomas or necrosis confirmed RDD. Initial management involved a three-week oral steroid regimen yielding transient improvement, though nodal recurrence necessitated therapeutic escalation. Methotrexate administration achieved sustained remission (Figure 5), with the patient remaining asymptomatic for 2 months and undergoing regular chest radiographic surveillance.

Figure 1: Child with massive bilateral cervical lymphadenopathy (before treatment).

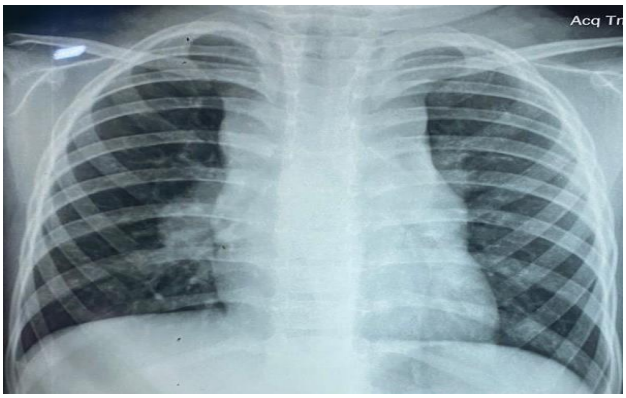

Figure 2: Plain skiagram of chest showing prominent hilar lymphadenopathy.

Figure 3: HRCT thorax showing mediastinal lymphadenopathy.

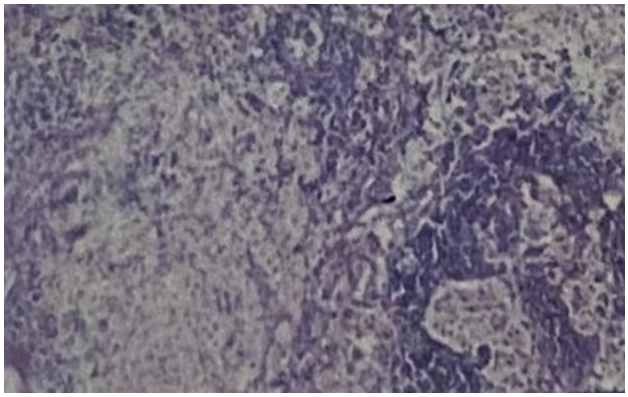

Figure 4: H & E-stained section of excised lymph node showing dense lymphohistiocytic proliferation with lymph node sinuses.

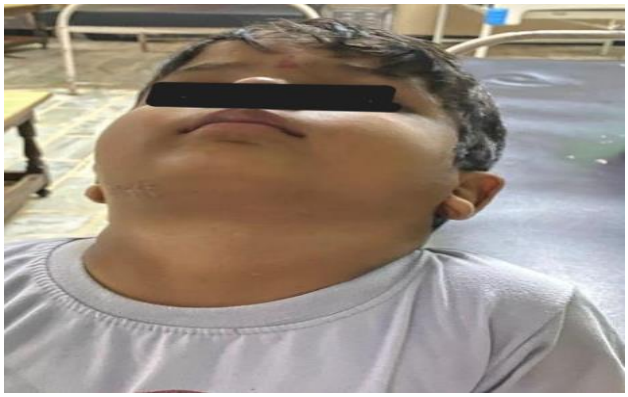

Figure 5: Child with no lymphadenopathy (after treatment).

DISCUSSION

RDD represents an uncommon histiocytic disorder first identified as 'sinus histiocytosis with massive lymphadenopathy' through seminal work by Destombes (1965) followed by Rosai and Dorfman (1969).[4,5] While etiology remains incompletely understood, kinase mutations exist and have been associated with nodal and extra nodal presentations excluding cutaneous forms. Classic manifestations include bilateral cervical lymphadenopathy, accompanied by fever, weight loss and night sweats, were partially observed here, with absence of systemic symptoms noted.[6] Diagnostic challenges emerge when concurrent nodal and extra nodal involvement occurs, necessitating histopathological confirmation as performed via cervical lymph node biopsy in this case.[7]

Clinical differentiation from IgG4 related disease proves critical due to overlapping features like storiform fibrosis.[6] Though spontaneous resolutions frequently occurs in sporadic cases, therapeutic strategies require tailored approaches based on anatomical involvement.[8] While isolated lymphadenopathy often warrants monitoring, vital organ compromise or obstructive symptoms may demand aggressive intervention. Mortality data underscores disease severity. Foucar et al (1990) documented 17

fatalities among 238 cases (7%), while Pulsoni et al (2022) reported 10 deaths in 80 patients (12%).[2,8] A critical consideration for differential diagnosis. Prognostic awareness remains essential when managing this clinically heterogenous condition. Treatment protocols incorporate corticosteroids (prednisolone 40-70 mg per day or dexamethasone 8-20 mg per day) alongside chemotherapeutic agents (methotrexate, vincristine, 6-mercaptopurine), low-dose interferon, antibiotic therapy, and selective radiation therapy. Node size and symptomatic presentation are frequently mitigated through steroid administration; though clinical outcomes demonstrate variability across patient populations. Clinicians occasionally prioritize chemotherapy as initial intervention for disseminated or high-mortality presentations, though this approach is typically reserved for cases demonstrating resistance, relapse patterns or steroid contraindications. Gaurav et al documented corticosteroid administration as primary treatment in 17 (27%) of 57 cases, with positive responses observed in 56% of these instances.[1]

Surgical interventions may involve partial or complete resection. Resection of solitary lesions in multifocal disease should be reserved for extensive presentations exhibiting end organ compromise or neurological deficits. When surgical intervention is contraindicated or alternative therapies remain unsuitable, radiation therapy may be employed. It can also be used for patients demonstrating persistent or recurrent symptoms post-resection of isolated pathology.

The current case demonstrated positive responsiveness to oral steroid therapy combined with methotrexate, achieving asymptomatic status maintained through 2 months of ongoing monitoring. Available evidence suggests RDD treatment responses exhibit marked variability, characterised by alternating remission and exacerbation phases.[3]

Follow up

The child showed positive response to oral steroid therapy combined with methotrexate within 2 months of treatment. He is being monitored at regular intervals for assessing response to therapy and management of the side effects of treatment through physical examination and with plain skiagram of the chest and has been asymptomatic.

CONCLUSION

Given the varying outcomes with similar histopathology, RDD's categorization as a syndrome, rather than a single disease entity, is often suggested. At one end of the spectrum, patients present with “benign” single-system unifocal RDD; an example is a solitary subcutaneous nodule which can be effectively observed or excised, potentially culminating in sustained remissions. Conversely, at the other end of the spectrum, are patients who exhibit a truly neoplastic condition. Close monitoring or specific systemic therapy is frequently warranted in such individuals. It requires a high degree of suspicion by the paediatricians and oncologists for timely diagnosis and intervention.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Goyal G, Ravindran A, Young JR, Shah MV, Bennani NN, Patnaik MM, et al. Clinicopathological features, treatment approaches, and outcomes in Rosai-Dorfman disease. Haematologica. 2019;105(2):348.
2. Foucar E, Rosai J, Dorfman R. Sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease): review of the entity. Semin Diagnost Pathol. 1990;7(1):19-73.
3. Singh A, Kanaujiya SK. The Rosai–Dorfman Disease: A Differential Diagnosis in Cervical Swelling. Indian J Otolaryngol Head Neck Surg. 2019;71(1):107-12.
4. Destombes P. Adenitis with lipid excess, in children or young adults, seen in the Antilles and in Mali.(4 cases). Bulletin de la Societe de pathologie exotique et de ses filiales. 1965;58(6):1169-75.
5. Rosai J. Sinus histiocytosis with massive lymphadenopathy. A newly recognized benign clinicopathological entity. Arch Pathol. 1969;87:63-70.
6. Bruce-Brand C, Schneider JW, Schubert P. Rosai-Dorfman disease: an overview. J Clin Pathol. 2020;73(11):697-705.
7. Abraham ZS, Francis Z, Ngaiza AI, Kahinga AA, Bukanu F. Histocytopathological diagnosis of Rosai–Dorfman disease: Case report. Clin Case Rep. 2022;10(2):e05412.
8. Pulsoni A, Anghel G, Falcucci P, Matera R, Pescarmona E, Ribersani M, et al. Treatment of sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease): report of a case and literature review. Am J Hematol. 2002;69(1):67-71.

Cite this article as: Goyal S, Sharma CM, Garg C. Rosai Dorfman disease: a rare entity. Int J Contemp Pediatr 2025;12:1226-8.