Clinical Case Reports

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Rosai-Dorfman Disease in a 4-Month-Old Female Presenting With Cervical Lymphadenopathy

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ABSTRACT

Rosai-Dorfman disease is a rare histiocytic disorder that can mimic malignancies. This case highlights the importance of immunohistochemistry in distinguishing RDD from lymphoproliferative neoplasms, ensuring accurate diagnosis and management. Clinicians should consider RDD in unexplained soft tissue masses to avoid misdiagnosis and unnecessary interventions.

1 | Case Presentation

A 4-month-old female of Pakistani origin presented with a progressively enlarging, non-tender swelling on the right side of the neck for 2 weeks. The parents denied fever, weight loss, night sweats, or other systemic symptoms. The patient was otherwise healthy, with no significant past medical history. There was no family history of such a condition, and her parents were not cousins.

On examination, there was a firm, mobile, 3 cm × 2 cm lymph node in the right cervical region, with no overlying skin changes or signs of inflammation. No other lymphadenopathy or organomegaly was detected. Laboratory investigations, including a complete blood count, were within normal limits. Viral serologies and tuberculosis screening tests were negative.

Ultrasound of the cervical region showed an enlarged, well-defined hypoechoic lymph node in the cervical region and no significant necrosis or abscess formation (Figure 1). X-ray chest demonstrated mediastinal widening and no bony structure involvement (Figure 2).

Fine needle aspiration cytology (FNAC) revealed large histiocytes exhibiting emperipolesis (engulfment of intact lymphocytes and plasma cells). An excisional biopsy of the lymph node was performed, and histopathology (Figure 3) demonstrated sinus histiocytosis with emperipolesis. Immunohistochemistry (Figure 4) confirmed the diagnosis, with the histiocytes staining positive for S-100, anti-CD68 antibodies, and cyclin D1 and negative for anti CD1a antibodies, excluding Langerhans cell histiocytosis. Cyclin D1 antibody testing was done to assess whether the histiocytic disorder has a mutation in the Map Kinase pathway. Special stains AFB and ZN were negative for fungal organisms and atypical mycobacteria.

The diagnosis of Rosai-Dorfman disease was made based on these findings. Given the absence of systemic symptoms or compressive effects, the patient was managed conservatively with close follow-up. At the 6-month follow-up, the lymphadenopathy showed significant regression.

Rosai-Dorfman disease is an extremely rare benign histiocytic proliferative disorder and has a prevalence of around 1 in 200,000 people [1]. It is commonly seen in children and young

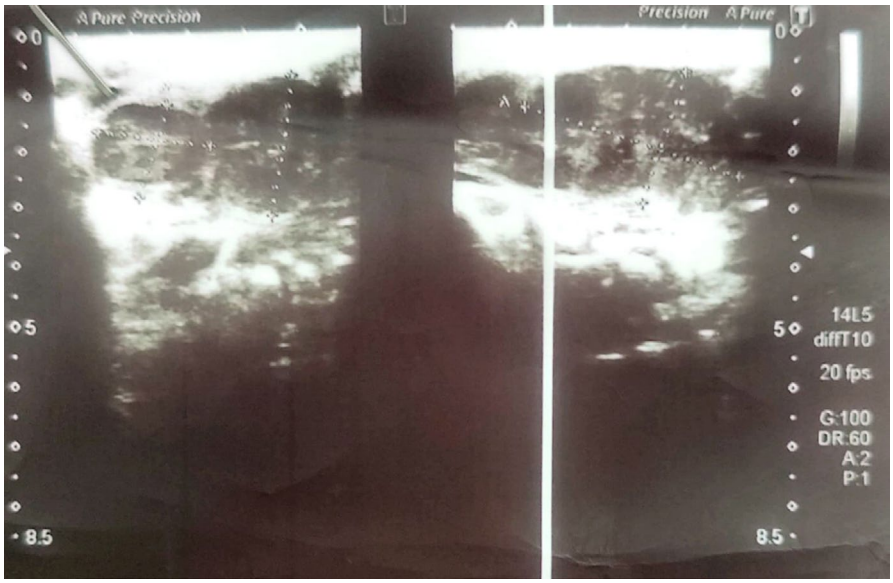

FIGURE 1 | Ultrasound showing an enlarged, well-defined hypoechoic lymph node in the cervical region. The lesion appears non-suppurative, with no signs of abscess formation or necrosis.

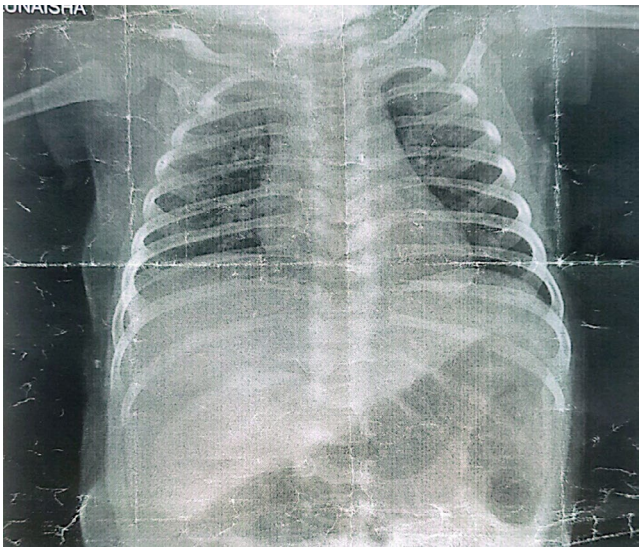

FIGURE 2 | X-ray chest demonstrating mediastinal widening, suggestive of lymphadenopathy. No lung parenchymal abnormalities, pleural effusion, or bony involvement are observed.

adults. The majority of diagnoses are typically made around the age of 20 [2]. Rosai-Dorfman disease is observed more frequently in males than in females [1]. It most frequently involves cervical lymph nodes, with systemic involvement in about 43% of cases [1]. The presence of plasma cells and characteristic immunohistochemical markers confirm the diagnosis. The clinical observation of emperipolesis in RDD remains common, but it does not qualify as a diagnostic requirement because it lacks strict specificity. The frequency of emperipolesis tends to be higher in nodal disease when compared to extranodal disease sites [3]. The histiocytes are positive for S100, fascin, CD68, and variable CD163 and CD14. Unlike Langerhans cell histiocytosis, the cells in Rosai-Dorfman disease are negative for CD1a and CD207 [1]. The Histiocyte Society advises quantifying IgG4-positive plasma cells, though results require careful interpretation. Any increase should be noted as an auxiliary finding, necessitating correlation with clinical, serological, and radiological data [3]. Management typically involves observation, with systemic therapy reserved for symptomatic or disseminated disease [1]. Sporadic RDD generally has a good prognosis, often resolving spontaneously in up to 50% of cases. However, around 10% of cases may result in death due to direct complications [3].

This case highlights the rarity of Rosai-Dorfman disease in a female infant, emphasizing the importance of cytological and histopathological evaluation in diagnosing uncommon pediatric lymphadenopathies, particularly in atypical age groups and genders.

Author Contributions

Allahdad Khan: conceptualization, methodology, project administration, supervision, writing – original draft, writing – review and editing. **Anam Malik:** conceptualization, data curation, investigation, writing – original draft. **Muhammad Hussnain Sadiq:** conceptualization, data curation, investigation, writing – original draft. **Muhammad Shahzaib Arshad:** conceptualization, data curation, investigation, writing – original draft. **Fatima Safdar:** conceptualization, investigation, writing – original draft. **Mohamed Antar:** conceptualization, project administration, writing – review and editing.

Consent

Written informed consent from the parents of the patient was taken for publication of this case and associated images.

Conflicts of Interest

The authors declare no conflicts of interest.

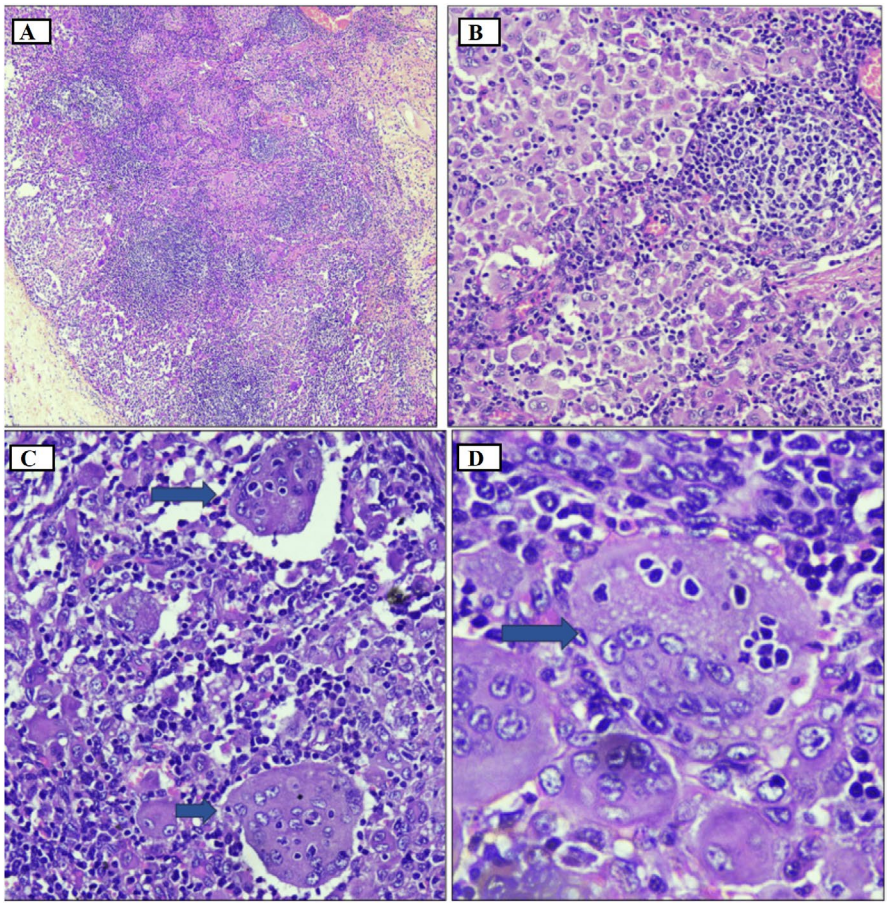

FIGURE 3 | (A) Low power view of lymph node with distended sinuses (H&E, ×10 magnification). (B) Sinuses exhibit histiocytes accumulation, with enlarged, round to oval nuclei and abundant eosinophilic cytoplasm (H&E, ×20 magnification). (C, D) Low and higher power views showing emperipolesis with engulfment of lymphocytes, respectively, marked by arrows (H&E, ×20, ×40 magnification).

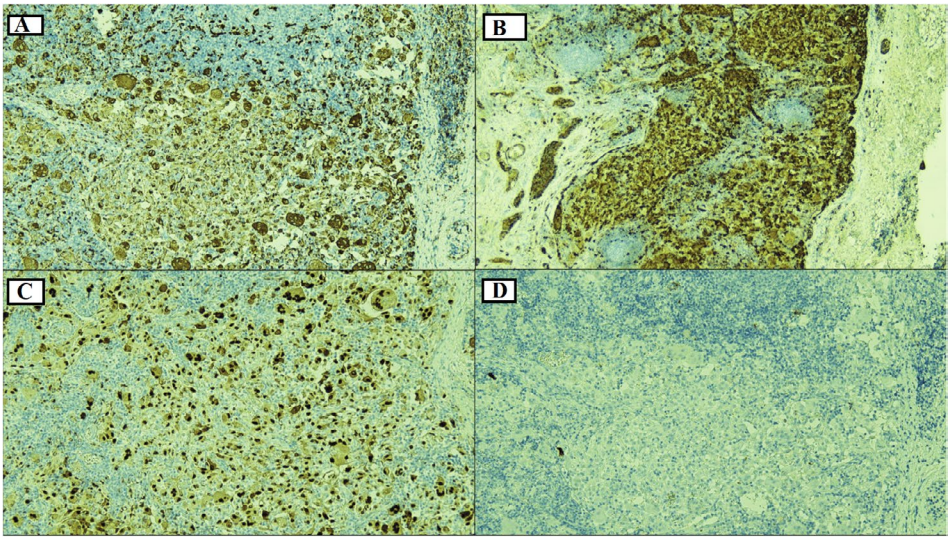

FIGURE 4 | (A) IHC immunostaining with anti-CD68 is positive in the histiocytic cells. (B) IHC immunostaining with anti-S100 is positive in the histiocytic cells. (C) IHC immunostaining with anti-Cyclin D1 is positive in the histiocytic cells. (D) IHC immunostaining with anti-CD1a is negative.

Data Availability Statement

Data available on request from the authors.

References

1. O. Abla, E. Jacobsen, J. Picarsic, et al., "Consensus Recommendations for the Diagnosis and Clinical Management of Rosai-Dorfman-Destombes Disease," *Blood* 131, no. 26 (2018): 2877–2890, https://doi.org/10.1182/blood-2018-03-839753.

2. Z. Kutlubay, O. Bairamov, A. Sevim, C. Demirkesen, and M. C. Mat, "Rosai-Dorfman Disease: A Case Report With Nodal and Cutaneous Involvement and Review of the Literature," *American Journal of Dermatopathology* 36, no. 4 (2014): 353–357, https://doi.org/10.1097/DAD.0b013e31829e5564.

3. T. Nguyen, M. Gutema, J. Ye, and M. S. Backenstoss, "Mammary Rosai-Dorfman Disease: Rare Benign Mimic of Breast Malignant Neoplasm," *Journal of Clinical Imaging Science* 13 (2023): 24, https://doi.org/10.25259/JCIS_40_2023.