

Genetic Basis of Thyroid Carcinoma: A Short Communication

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Abstract

Background

Rosai-Dorfman disease (RDD) is a rare, benign disorder characterized by painless, massive cervical lymphadenopathy. First described in 1965, it commonly presents with fever, leukocytosis, elevated ESR, and hypergammaglobulinemia. Diagnosis is confirmed by histopathology showing histiocyte proliferation with emperipolesis and positive S-100 and CD68 staining.

Case Presentation

A 12-year-old girl presented with a gradually enlarging left cervical lymph node. Fine-needle aspiration cytology and excisional biopsy confirmed RDD. After surgical excision, she was treated with steroids, leading to regression of lymphadenopathy.

Conclusions

RDD generally has a benign, self-limiting course. However, careful monitoring is essential due to possible relapse or extranodal involvement. Early diagnosis and appropriate management yield favorable outcomes.

Keywords: Case Report, Histiocytosis, Juvenile, Lymph Nodes, Rosai-Dorfman Disease, Sinus Histiocytosis.

Introduction

Rosai-Dorfman disease (RDD), or sinus histiocytosis with massive lymphadenopathy (SHML), is a benign, self-limiting disorder marked by painless cervical lymph node enlargement¹. Classic symptoms include fever, leukocytosis, and hypergammaglobulinemia². Notably, 40% of cases exhibit extranodal involvement in organs such as skin, eyes, nervous system, bone, gastrointestinal tract, and more, often mimicking other conditions²⁻⁶. Diagnosis of nodal RDD is typically straightforward on

H&E staining with supportive immunohistochemistry, but extranodal presentations or those with fibrosis and minimal emperipolesis can be challenging⁷. Our case is notable for presenting as isolated unilateral cervical lymphadenopathy in a 12-year-old without systemic symptoms or extranodal involvement. FNAC followed by excisional biopsy confirmed RDD, highlighting the importance of histological evaluation even in clinically ambiguous cases. The rarity of such isolated pediatric presentations without systemic signs makes this case

unique and worthy of reporting.

Case Report

12-year-old female presented to the Otorhinolaryngology outpatient department of Dhulikhel Hospital with a one-year history of swelling over the left upper lateral aspect of the neck (Figure 1). The swelling was painless and gradually progressive, with no associated history of fever or cough.



Figure 1. *Rosai-Dorfman disease: A 12-year-old girl with left cervical lymphadenopathy*

Clinical examination revealed multiple enlarged left cervical lymph nodes, ranging in size from 1×2 cm to 2×2 cm. The overlying skin appeared normal, with no change in color or local rise in temperature. The lymph nodes were non-tender, firm to hard in consistency, mobile, with smooth and regular surfaces, and not adherent to the skin. No other lymphadenopathy was detected.

Ultrasonography of the neck showed multiple enlarged lymph nodes with loss of central fatty hilum at levels IB and V on the left side, the largest measuring 25×11 mm. CT scan of the neck revealed multiple homogeneously enhancing, discrete enlarged lymph nodes in the left cervical region (levels IB and V), the largest measuring 25×22 mm.

Fine-needle aspiration cytology (FNAC) of the left cervical lymph nodes showed numerous histiocytes with eccentrically placed round-to-oval nuclei, prominent nucleoli, and abundant pale cytoplasm, with evidence of lymphophagocytosis (emperipolesis). The cytological diagnosis was Rosai-Dorfman disease (RDD). The patient was initially managed with anti-inflammatory medication and kept under regular follow-up. However, after two months, the swelling did not regress, and an excisional biopsy was planned. Under general anaesthesia, a single soft, yellowish, well-circumscribed encapsulated globular lymph node mass measuring 3×3 cm was excised (Figure 2).



Figure 2. *Photograph of excised lymph node*

Histological examination revealed encapsulated lymphoid tissue with effaced architecture (Figure 3). The distended sinuses contained a prominent population of histiocytes characterized by large round-to-oval nuclei, prominent nucleoli, and abundant cytoplasm showing emperipolesis, engulfment of intact lymphocytes.

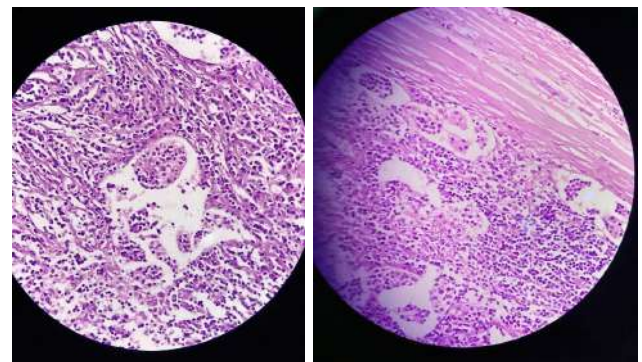


Figure 3. *Photomicrograph showing lymphoid tissue with effaced architecture and emperipolesis, suggestive of sinus histiocytosis*

The intervening stroma showed an increased number of plasma cells, blood vessels, and areas of haemorrhage. The final diagnosis was sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease). Postoperatively, the patient was treated with oral corticosteroids for two weeks, after which progressive regression of the lymph node swelling was observed (Figure 4).



Figure 4. *Post-operative photograph after excision of the lymph nodes*
The patient remains under regular follow-up, with no evidence of recurrence to date.

Conclusion

Rosai-Dorfman disease (RDD) is a rare, benign histiocytic disorder that can mimic more serious pathologies, particularly when presenting as isolated lymphadenopathy without systemic symptoms. Early

recognition based on characteristic cytological features especially emperipolesis, supported by histopathology and immunohistochemistry, is essential for accurate diagnosis and appropriate management. Our case highlights an uncommon presentation of RDD as unilateral cervical lymphadenopathy in a pediatric patient without systemic or extranodal involvement, which makes it clinically unique and diagnostically challenging. Surgical excision combined with a short course of corticosteroids led to complete resolution. This case reinforces the importance of considering RDD in the differential diagnosis of persistent cervical lymphadenopathy in children and the value of a multidisciplinary approach in ensuring effective management and follow-up.

Consent

Written informed consent was obtained ensuring patient's anonymity.

Declaration of competing interest

There are no conflicts of interest.

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N/A

References

1. Deshpande V, Verma K. Fine needle aspiration (FNA) cytology of Rosai-Dorfman disease. *Cytopathology*. 1998;9(5):329–35.
2. Park YK, Kim YW, Choi WS, Lim YJ. Sinus histiocytosis with massive lymphadenopathy: Multiple skull involvement. *J Korean Med Sci*. 1998;13(4):423–7.
3. Hummel P, Waisman J, Chhieng D, et al. Fine-needle aspiration cytology of Rosai-Dorfman disease of the breast: A case report. *Diagn Cytopathol*. 1999;21(4):287–91.
4. Kim M, Provias J. Rosai-Dorfman disease mimicking multiple meningioma: Case report. *Neurosurgery*. 1995;36(6):1185–7.
5. Foucar E, Rosai J, Dorfman R. Sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease): Review of the entity. *Semin Diagn Pathol*. 1990;7(1):19–73.
6. Rosai J, Dorfman RF. Sinus histiocytosis with massive lymphadenopathy: A pseudolymphomatous benign disorder. Analysis of 34 cases. *Cancer*. 1972;30(5):1174–88.
7. Bruce-Brand C, Schneider JW, Schubert P. Rosai-Dorfman disease: An overview. *J Clin Pathol*. 2020;73(11):697–705.
8. Abraham ZS, Francis Z, Ngaiza AI, Kahinga AA, Bukanu F. Histocytopathological diagnosis of Rosai–Dorfman disease: Case report. *Clin Case Rep*. 2022;10(8):e05412.
9. Werneck Rodrigues DO, Wolp Diniz R, Dentz LC, Costa MA, Lopes RH, Suassuna LF, et al. Case study: Rosai-Dorfman disease and its multifaceted aspects. *J Blood Med*. 2024;15:123–8.
10. Rastogi V, Sharma R, Misra SR, Yadav L, Sharma V. Emperipolesis – A review. *J Clin Diagn Res*. 2014;8(12):ZM01–2.
11. Amaraweera P, Perera HA. A case of Rosai-Dorfman disease. *Galle Med J*. 2009;11(2):49–50.
12. Danisious T, Hettiarachchi M, Dharmadasa C, Jayaweera H. Rosai-Dorfman disease with renal involvement and associated autoimmune haemolytic anaemia in a 12-year-old girl: A case report. *BMC Pediatr*. 2020;20(1):1–5.
13. Biswal P, Dhal I, Kar A, Nayak M, Pradhan D, Naik S. Diagnosis of multifocal extranodal Rosai-Dorfman disease by fine needle aspiration cytology. *J Adv Med Med Res*. 2015;3(9):872–6.
14. Ashish G, Chandrashekharan R, Parmar H. Rare case of Rosai-Dorfman disease involving paranasal sinuses in paediatric patient: A case report. *Egypt J Ear Nose Throat Allied Sci*. 2016;17(1):43–6.
15. Jabali Y, Smrcka V, Pradna J. Rosai-Dorfman disease: Successful long-term results by combination chemotherapy with prednisone, 6-mercaptopurine, methotrexate, and vinblastine: A case report. *Int J Surg Pathol*. 2005;13(3):285–9.