Radiology Case Reports 19 (2024) 956–960

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Case Report

# Uncommon presentation of Rosai-Dorfman disease: Nasal and nasopharyngeal involvement: A case report and discussion☆

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ARTICLE INFO

*Article history:*
Received 8 September 2023
Revised 18 November 2023
Accepted 22 November 2023

*Keywords:*
Rosai-Dorfman disease
Extranodal presentation
Case report
Immunohistochemistry

ABSTRACT

This study presents a rare case of Rosai-Dorfman disease (RDD) with nasal and nasopharyngeal involvement, illustrating the complexities in diagnosing this enigmatic histiocytic disorder. RDD, characterized by massive, painless cervical lymphadenopathy, poses diagnostic challenges due to its diverse clinical presentations. In this case, a 38-year-old woman presented with a year-long history of neck swellings, nasal congestion, headaches, and sinusitis-like symptoms. Radiological imaging and histopathological examination revealed RDD involvement in the nasopharynx and paranasal sinuses. RDD diagnosis was confirmed through immunohistochemistry. The patient's unique symptoms emphasize the importance of considering RDD in the differential diagnosis of sinonasal masses with recurrent or unusual complaints. This case underscores the need for increased awareness, multidisciplinary management, and further research to enhance understanding and treatment of RDD, especially in extranodal presentations.



## Introduction

Rosai-Dorfman disease (RDD), also known as sinus histiocytosis with massive lymphadenopathy (SHML), is a rare and enigmatic histiocytic disorder first described by Destombes in 1965 and subsequently characterized by Rosai and Dorfman in 1969 [1,2]. This intriguing disease presents a wide spectrum of clinical manifestations and outcomes, making it a subject of significant medical interest. RDD is classified within the non-

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2023.11.055

Langerhans cell histiocytosis group, and according to recent classification, it falls under the "R group" histiocytosis [3].

The hallmark of RDD is often the massive, painless, and bilateral cervical lymphadenopathy, accompanied by fever, leukocytosis, elevated erythrocyte sedimentation rate, and polyclonal hypergammaglobulinemia [4]. While this lymphadenopathy is the most common presentation, RDD can also manifest as an extranodal disease affecting various sites, including the skin, soft tissue, central nervous system, gastrointestinal tract, breast, and sino-nasal region [3].

The diagnosis of RDD primarily relies on histopathological examination, where a characteristic feature called "emperipolesis," which involves histiocytes engulfing lymphocytes and other blood cells, is observed. This unique histopathological feature, along with immunohistochemical markers, aids in confirming the diagnosis [5].

Given the diversity in clinical presentations, RDD poses diagnostic challenges, often mimicking malignancies or infectious diseases [2,6]. Understanding the nature of RDD, its varied clinical course, and the potential for spontaneous remission or complications is essential for timely and appropriate management [6]. In this context, we present a case of RDD with nasal and nasopharyngeal involvement, emphasizing the complexity and diagnostic considerations associated with this rare disease. This case highlights the importance of raising awareness and further research to bridge the knowledge gap surrounding RDD.

## Case presentation

A 38-year-old woman visited our outpatient department with a complaint of multiple neck swellings that have been present for the past year. Initially, the patient noticed a swelling on her left neck, roughly the size of a pea, which gradually grew and eventually led to bilateral neck swellings. Despite their growth, the swellings were not accompanied by pain or any bothersome symptoms, which is why she did not seek medical advice. However, over time, she began to notice similar swellings in other areas of her body, such as her cheeks and the front of her chest. Alongside these swellings, she developed nasal congestion and began experiencing recurrent headaches and sinusitis. Furthermore, she had 2 episodes of nosebleeds in the last 6 months, both of which resolved without requiring any medical intervention. Additionally, she had been dealing with difficulty swallowing for the past 6 months, although it was not accompanied by pain during swallowing. The patient had experienced intermittent fever over the past year, although she did not document her temperature. Notably, she did not report any weight loss, changes in appetite, abnormal body movements, loss of consciousness, cough, chest pain, shortness of breath, night sweats, alterations in bowel or bladder habits, or changes in voice or vision. Her systemic review revealed no remarkable findings, and she neither smoked nor consumed alcohol. The patient had no noteworthy past medical history, and her family history did not include any chronic diseases. She had never undergone any surgical procedures and had no known allergies. Neither the patient nor any close contacts had ever received a diagnosis or treatment for tuberculosis. She was sexually active with her husband, with whom she had been married for the past 10 years.

Upon examination, the patient exhibited overall good health, had a moderate build, and displayed appropriate orientation to time, place, and person. Her BMI was calculated at 22.3 kg/m$^2$. At the time of presentation, she did not have a fever and had a blood pressure reading of 110/70 mm Hg, a pulse rate of 70 beats per minute with a regular rhythm, a respiratory rate of 16 breaths per minute, and maintained a room air O2 saturation level of 95%. She showed mild pallor but exhibited no signs of jaundice or dehydration. During the examination, multiple enlarged lymph nodes were palpable on both sides, situated anteriorly and posteriorly to the sternocleidomastoid muscle, with the largest one measuring approximately 2.5 × 2.5 cm. These lymph nodes had a smooth texture, felt firm to the touch, were freely movable, and did not elicit tenderness upon palpation. There were no observable skin changes or pulsations over them. An oral examination revealed bilaterally enlarged tonsils without the presence of exudates. A systemic examination did not reveal any noteworthy findings. A nasopharyngolaryngoscopy (NPL) was conducted, which indicated adenotonsillar hypertrophy along with bilateral nasal polyps.

Based on the history provided and the findings from the examination, the patient was suspected to have lymphoma. To further investigate, a series of tests and imaging studies were conducted, including a complete blood count, erythrocyte sedimentation rate (ESR), renal function test, serum electrolytes, chest X-ray, and magnetic resonance imaging (MRI) of the neck. The results showed elevated ESR levels, and the MRI revealed various significant findings. These included lobulated iso signal lesion in the right nasal cavity and enlarged nasopharyngeal tonsil (Fig. 1), which shows homogenous enhancement in postcontrast images (Figs. 2A and B). Homogenous enlargement of bilateral palatine tonsils and bilateral cervical lymphadenopathy is also noted (Fig. 2C). Subsequently, a biopsy was performed on a lymph node at level II.

Histopathology analysis of the lymph node showed dilated sinuses with partial effacement of the lymphocytes, plasma cells, and histiocytes, some of these histiocytes having numerous intact lymphocytes in the cytoplasm suggesting emperipolesis. These findings indicated sinus histiocytosis with massive lymphadenopathy, suggesting the need for immunohistochemistry. Despite facing financial constraints, the patient consented to the necessary investigations to establish a diagnosis. Subsequently, the tissue sample was sent for further examination. Immunohistochemistry analysis revealed the presence of CD20-positive lymphoid follicles and the expression of CD68 and CD3 by foamy histiocytes, ultimately establishing a conclusive diagnosis of sinus histiocytosis with features consistent with RDD. Additional imaging studies were conducted to eliminate the possibility of internal organ involvement. The patient was informed about the rare nature of the disease and received clear counseling regarding its nonmalignant nature at present. Given that this condition typically undergoes spontaneous remission in most patients, the patient was recommended to maintain regular follow-up appointments for symptom monitoring. She underwent eval-

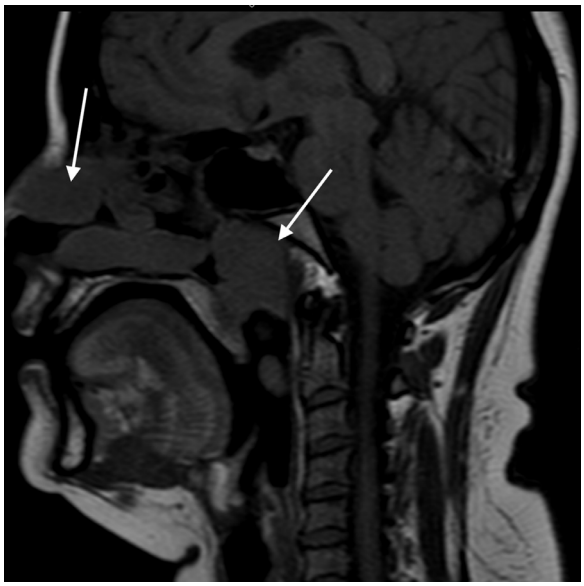

**Fig. 1 – Mid-sagittal T1-weighted magnetic resonance imaging shows iso signal intensity lobulated enlarged nasopharyngeal tonsil and another lesion in the right nasal cavity (white arrows).**

uations every 2 months; however, her symptoms did not remit even after 2 years. Hence, she was started on oral Prednisolone 1 mg/kg after which her symptoms started to regress significantly. After 2 weeks of the initial dose, the patient was continued on a reduced dose of 0.5 mg/kg. The swellings started to subside and the patient was in remission after treatment for 6 weeks. The patient was explained well about the possibility of relapse and is continuing regular follow-up visits.

## Discussion

The case described here demonstrates how RDD can present as well as the difficulties encountered when diagnosing this uncommon histiocytic disorder.

RDD, also known as sinus histiocytosis with massive lymphadenopathy, is an exceedingly rare histiocytic disorder with a broad spectrum of clinical presentations [1]. In this case, we presented a 3-year-old female with RDD, demonstrating the complexity of the disease. RDD can impact people of all ages, but it is most frequently seen in individuals in their twenties or thirties. It is more prevalent in males and those of African descent. RDD can affect individuals across different age groups and can show up in various organ systems, affecting both children and adults. Additionally, there is a higher occurrence in males among the adult population [3,7,8]. Common clinical features include painless lymphadenopathy, fever, night sweats, and weight loss [9]. However, RDD's clinical presentation is highly variable, and as evidenced in this case, it can involve the nasopharynx, leading to unique symptoms. RDD primarily involves lymph nodes but can manifest as extranodal involvement in various anatomical sites, including the head and neck region, central nervous system, skin, soft tissues, and more [3,6]. In this case, the patient exhibited an extranodal involvement in the nasal cavity and paranasal sinuses which is relatively uncommon in RDD cases [1].

In the study by Gaitonde et al. [10], the SHML registry, comprising 423 cases, indicates that there is an equal prevalence of the disease among people of African descent and whites. Furthermore, it suggests that individuals of Asian descent are less commonly affected by this condition which is notable while describing the presentation of the disease in an Asian male patient in the presented case.

Radiological imaging plays a pivotal role in the diagnosis and assessment of RDD. The MRI scan in this case revealed an

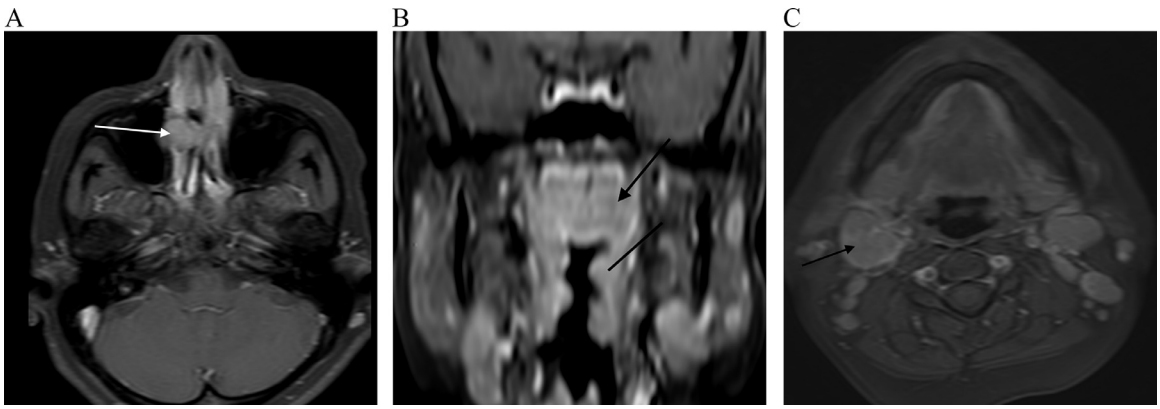


**Fig. 2 – (A) Axial T1-weighted postcontrast magnetic resonance imaging showing homogenous enhancing lesion (white arrow) in the right nasal cavity causing mild displacement of the nasal septum to the contralateral side. (B) Mid-coronal T1-weighted postcontrast magnetic resonance imaging showing the enlarged homogenously enhancing nasopharyngeal tonsil (black arrow) and palatine tonsil (black line) causing mild luminal narrowing. (C) Axial postcontrast magnetic resonance imaging showing the enlarged bilateral homogenously enhancing lymph nodes largest in the right level II region (black arrow).**

enhancing lesion in the nasopharynx measuring 19 × 18 × 17 mm. This finding highlights the importance of radiological investigations in identifying the extent and location of RDD lesions.

RDD can present as single or multiple masses in various anatomical sites, making imaging crucial for accurate diagnosis and treatment planning. In this case, the presentation in the nasal cavity and paranasal sinuses is less commonly documented in the literature, making the diagnosis challenging due to its rarity [1,3,7].

Definitive diagnosis of RDD relies on histopathological examination [6]. Biopsy of the nasopharyngeal lesion in our case provided the conclusive evidence needed to confirm RDD. Histologically, RDD is characterized by the presence of histiocytes with emperipolesis, a hallmark feature where intact lymphocytes are engulfed within histiocytic cytoplasm which differentiates it from other histiocytic disorders such as chronic inflammation, rhinoscleroma, Erdheim-Chester disease, and Langerhans cell histiocytosis Immunohistochemistry can further confirm RDD, with positive staining for S100 protein and CD68 [7,11]. Accurate diagnosis is essential to distinguish RDD from other conditions with similar clinical presentations.

The patient's history of nasal blockage and recurrent sinusitis is noteworthy. RDD involving the nasopharynx can lead to obstructive symptoms and sinusitis-like complaints. These symptoms, although nonspecific, emphasize the importance of considering RDD in the differential diagnosis of sinonasal masses, especially in cases where recurrent or atypical symptoms are observed [1,11].

The presence of adenoid or tonsillar hypertrophy in this case raises intriguing questions about potential associations between RDD and lymphoid tissue. Further research is needed to elucidate the underlying mechanisms and clinical implications of such associations.

Treatment of RDD remains challenging due to its rarity and variable clinical course. While in most patients with uncomplicated lymphadenopathy, observation is crucial due to its risk of remission, surgery, corticosteroids, immunomodulatory therapy, targeted therapies, and radiotherapies are some of the treatment options available [3,7]. In this case, the patient received targeted therapy with Rituximab, which has shown promise in some RDD cases [12]. However, the optimal treatment approach for RDD remains uncertain, and further studies are needed to establish standardized guidelines.

RDD poses diagnostic challenges, often masquerading as malignancies or infectious diseases due to its variable clinical presentation. A thorough evaluation and appropriate diagnostic tests are essential to exclude other potential diagnoses [2]. It is crucial for healthcare providers to maintain a high index of suspicion and consider RDD in the differential diagnosis, especially in cases with unusual clinical findings.

RDD's diverse clinical manifestations underscore the need for a multidisciplinary approach to diagnosis and management [7]. Understanding the spectrum of RDD presentations is vital for timely and accurate patient care.

The prognosis of RDD varies, with some cases demonstrating spontaneous remission, while others follow a more unpredictable course. Long-term follow-up is necessary to monitor disease progression and response to treatment. Outcomes are usually favorable, particularly for cases of nodal and cutaneous disease, which are often self-limited [6,7].

Raising awareness about RDD is critical among healthcare providers and the medical community to enhance early recognition and accurate diagnosis. Improved awareness can lead to timely intervention and better patient outcomes [2].

Given the rarity of RDD, further research is needed to better understand the etiology, pathogenesis, and optimal treatment approaches for RDD, particularly in extranodal presentations. Additionally, raising awareness and sharing rare cases like this one can contribute to expanding the knowledge base about RDD.

## Conclusion

This case highlights the extranodal presentation of RDD with nasal and nasopharyngeal involvement. RDD's clinical diversity poses diagnostic challenges, emphasizing the importance of considering RDD in the differential diagnosis of sinonasal masses with recurrent or unusual symptoms. Multidisciplinary collaboration, including radiological imaging and histopathological analysis, is essential for accurate diagnosis. While RDD's optimal treatment remains uncertain, early intervention can lead to favorable outcomes. This case emphasizes the need for increased awareness, research, and standardized guidelines to enhance RDD's understanding and management, particularly in extranodal manifestations.

## Patient consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

## Ethical approval

This case report did not require review by the ethical committee.

## Author contributions

**Saubhagya Dhakal:** Conceptualization, as mentor and reviewer for this case report and for data interpretation. **Shailendra Katwal:** Contributed to performing literature review and editing. **Aastha Ghimire and Amrit Bhusal:** Contributed to writing the paper and reviewer for this case. **Tek Nath Yogi:** Contributed to writing the paper. All authors have read and approved the manuscript.

## Registration of research studies

Not applicable.

## Provenance and peer review

Not commissioned, externally peer-reviewed.

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