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CASE REPORT

A Challenging Rare Case: Rosai-Dorfman Disease Presented as a Single Osseus Lesion in a 16-Month-Old Boy

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Abstract

Sinus histiocytosis with massive lymphadenopathy (also known as Rosai-Dorfman disease (RDD)), a rare non-Langerhans cell histiocytosis, was originally recognized in 1969 by Rosai and Dorfman. RDD is characterized by the accumulation of activated histiocytes in various tissues and organs, but most commonly in lymph nodes. RDD is subclassified in two forms. The more common form, nodal RDD, and the rare form, extranodal RDD, which is based on the presence of extranodal tissue involvement. Most extranodal RDDs also involve lymph nodes. Primary, exclusively extranodal RDD is rare, and is exceedingly uncommon in young pediatric patients. These cases have rarely been reported in the current medical literature. In this case report, we present the diagnostic work-up of an osseous RDD in a 16-month-old male. This case reminds us that rare diseases with atypical presentation can happen in young children. Raising the awareness of rare diseases with unusual clinical presentation will help improve diagnostic accuracy and contribute to the effective management of these patients.

Keywords

Rosai-Dorfman disease, Bone lesion, Pediatric patients

Abbreviations

RDD: Rosai-Dorfman Disease; SHML: Sinus Histiocytosis with Massive Lymphadenopathy; LCH: Langerhans Cell Histiocytosis; ESR: Erythrocyte Sedimentation Rate; MRI: Magnetic Resonance Imaging; CT: Computerized Tomography

Introduction

Rosai-Dorfman Disease (RDD) was first described in 1969 as sinus histiocytosis with massive lymphadenopathy (SHML) by Rosai and Dorfman [1]. It is a rare, non-Langerhans cell histiocytosis with a prevalence of 1:200,000 and an estimated 100 new cases per year in the United States [2]. It is more commonly seen in children and young adults with a mean age of onset of 21 years. In 2016, the Histiocyte Society reclassified RDD into the "R" group of Histiocytoses, with further subclassification into Familial, Classical or Nodal, Extranodal, Neoplasia-associated, and Immune Disease-associated RDD [2]. The presence of large histiocytic cells with emperipolesis is the characteristic pathological feature. These cells stain positive for CD14, HLA-DR, CD68, CD163, S-100, and fascin, and negative for CD1a, differentiating RDD from Langerhans cell histiocytosis (LCH) [2]. The clinical presentation of RDD is usually subtle and non-specific. The most common initial presentation in patients with nodal RDD is painless, bilateral lymphadenopathy, most common cervical, accompanied by fever and weight loss [3]. Extranodal RDD may present either with a primary extranodal manifestation, or in conjunction with nodal RDD. Laboratory results are variable and nonspecific. Common findings range from leukocytosis, neutrophilia,

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and elevated erythrocyte sedimentation rate (ESR), to hypergammaglobulinemia [3].

In this paper, we report a case of a 16-month-old male with an unusual presentation of osseous RDD localized to the right distal radius, without lymph node or any other organ involvement.

Case Presentation

This 16-month-old male patient presented to the emergency department for a chief complaint of distal, right forearm pain, following a mild trauma to the arm while playing with a sibling. He was born at full term and was healthy prior to this incident. X-ray of the right forearm revealed a lytic lesion with a wide zone of transition, cortical destruction, and poorly defined periosteal reaction in the distal radius, which was concerning for an aggressive primary bone lesion (Figure 1A). Subsequent magnetic resonance imaging (MRI) of the right arm confirmed a destructive lesion in distal radius with the presence of a prominent adjacent soft tissue component. The imaging evaluation favored an aggressive bone lesion, such as Ewing sarcoma, given the patient's age. Laboratory results were normal for leukocyte counts, ESR, and immunoglobulins.

A needle core biopsy was performed for the diagnosis. The biopsy material was scant and fragmented. It was composed of reactive lamella bone with acute and chronic inflammatory infiltration, which was composed of neutrophils, lymphocytes, plasma cells and histiocytes. There was no evidence for malignancy. Based on scant material, it was suspicious of osteomyelitis. The patient received cephalexin for five days. Subsequently he underwent curettage with washout for the management of presumed osteomyelitis and to obtain more material for diagnosis. The curettage material showed mixed inflammatory infiltration with a prominent population of large histiocytic cells containing abundant foamy cytoplasm and showing emperipolesis (Figure 1B). Immunohistochemistry revealed that the histiocytic cells are positive for CD68, S-100 (Figure 1C), and negative for CD1a. The fungal, mycobacterial, and bacterial cultures, as well as PCR for *Kingella kingae*, were negative. A diagnosis of extranodal osseous RDD was rendered. An external expert bone pathology consultant confirmed the diagnosis.

The patient was staged with imaging studies, which confirmed the isolated right radius bone lesion without enlarged lymph nodes or any other organ or tissue involvement. Since the curettage removed the vast majority of lesional tissue, he received observation management with surveillance imaging. Follow-up computerized tomography (CT) scans of the right forearm up to 24 months following the diagnosis, showed bone sclerosis and remodeling, which is consistent with callus formation without a recurrent disease.

Discussion

The pathogenesis of RDD is not well-defined, and is likely variable based on the phenotypes. RDD is a non-neoplastic disease. Although recent studies identified gene mutations in patients with RDD, none could prove the neoplastic nature of RDD cells [4]. Rare familial cases were reported, but our patient does not have a family history of predisposing to RDD.

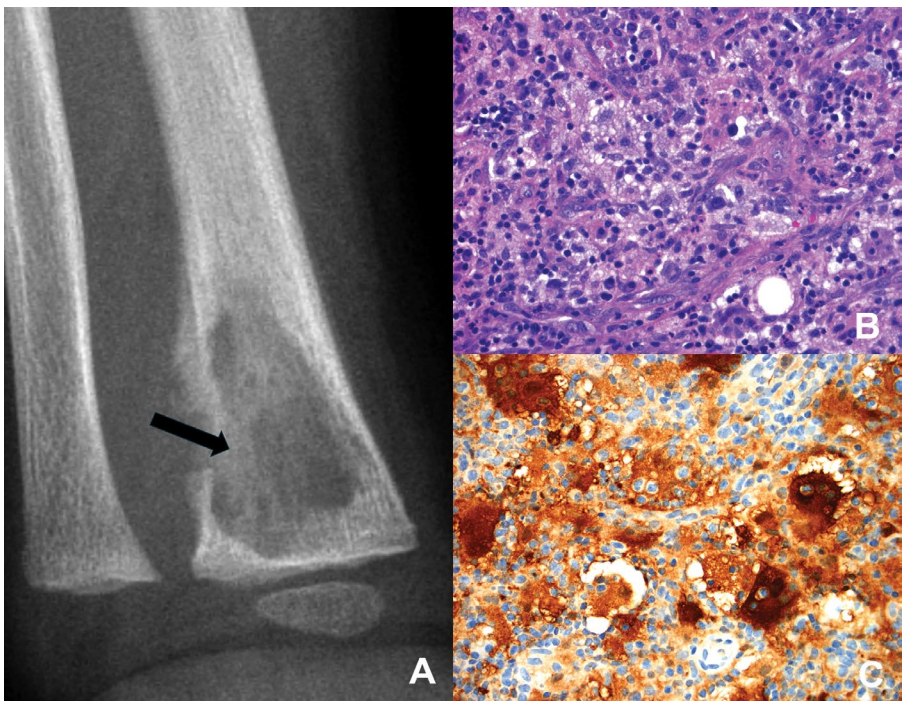

Figure 1: (A) X-ray of right radius revealed a lytic lesion with a wide zone of transition, cortical destruction (arrows), and poorly defined periosteal reaction; (B) Curettage specimen showed a mixed inflammatory infiltration including large foamy cells with emperipolesis (H&E, x40); (C) Most histiocytes stained strongly positive for S-100 (S-100 immunohistochemistry stain, x40).

Most patients with RDD presented with enlarged lymph nodes. Up to 40% of patients with RDD manifested extranodal involvement, with or without coexisting lymphadenopathy [5]. The most common extranodal site is skin and soft tissue [5]. Bone involvement occurs in 5% to 10% of RDD cases but is usually associated with or preceded by lymph node manifestations [6]. RDD with isolated bone involvement occurs in only 2% of patients [7]. The most common bone manifestations are cranium, facial bone and tibia [8]. RDD affects metaphysis or diaphysis of long bone with features of osteolytic process. The radiographic and histologic differential diagnosis is broad, ranging from chronic osteomyelitis, to lymphoma, and Ewing sarcoma. When a patient presents with bone RDD without lymph node involvement, it is important to keep the rare disease aspect of RDD in mind, otherwise it can be challenging to make the correct diagnosis. Recently, there were reports in two adult female patients with isolated osseus RDD. Both cases were misdiagnosed as chronic inflammation clinically and radiographically [9,10]. One of which was misdiagnosed histologically, based on small biopsy material [10].

Among the rare presentations of RDD exclusively involving bone, only 15 pediatric cases have been reported in the existing literature [7]. The age range of these pediatric patients was broad, though most cases involved children 10 years of age or older. The reports of young patients with isolated osseous RDD are exceedingly rare. In 1998, it was reported that a 2-year-old child presented with bone lesions in the left cuboid and right calcaneus. An excisional biopsy of cuboid revealed osseus RDD. But further work up discovered hilar lymph node involvement [11]. Similar to our case, in the recent literature a 17-month-old male, who had talus RDD without lymph node involvement was described [12]. The diagnosis was made on the curettage specimen and the bone healed one year after the surgery. The current case is likely the second case of RDD with isolated bone involvement in a child younger than 2 years of age. The addition of this case to the existing literature will raise the awareness of RDD with unusual clinical presentation in young patients. Clinicians of all specialties should keep extranodal RDD in mind to make the correct diagnosis in managing these patients, especially in the pediatric population.

Conclusion

Our case serves as a reminder that while osseous RDD in children is exceedingly rare, it does exist. Clinically, radiographically, and histologically, osseus RDD can mimic common bone diseases in children such as osteomyelitis, LCH, and Ewing sarcoma. Despite the diagnostic challenge, keeping RDD in the differential diagnosis can lead to appropriate work up and management, especially in young patients. A greater understanding of RDD, the pathophysiology, and risk factors could expedite the diagnosis of RDD for patients.

Source of Support

None.

Authors Contribution

All authors contributed to writing and prof editing of the manuscript, and providing important clinical, radiological and histological information.

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