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Rosai-Dorfman's disease, an uncommon cause of common clinical presentation

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CASE REPORT

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Abstract

Sinus histiocytosis with massive lymphadenopathy also known as Rosai-Dorfman disease is a rare benign disease that typically manifests as lymphadenopathy with or without systemic manifestations whose etiology remains poorly understood. Most common clinical presentation is painless bilateral cervical lymphadenopathy. However, it also can present in various extranodal sites and can easily be missed because of its rarity if not considered in the differential diagnosis. It commonly occurs in children and young adults with a slightly male predominance. Clinically patients may be mistaken for lymphoma and other infectious disorders like tuberculosis especially in developing countries like in Pakistan where tuberculosis is more prevalent. Here we report a case of a 38-years-old gentleman with bilateral cervical lymphadenopathy and worsening ascites. His symptoms initially mimicking tuberculosis and lymphoma, was finally diagnosed as having Rosai-Dorfman's Disease on intra abdominal lymph node biopsy.

Keywords: Sinus histiocytosis with massive lymphadenopathy, Rosai-Dorfman disease, Ascites.

Introduction

Rosai-Dorfman disease (RDD), also known as sinus histiocytosis with massive lymphadenopathy, is a rare, benign, non-Langerhanscell, histiocytic proliferative disease without any known cause.[1] Histopathological hallmark of Rosai-Dorfman disease is characteristic histiocytes with abundant pale cytoplasm exhibiting emperipolesis. Immunohistochemically strongly positive for S100 protein and various markers especially CD68 and CD163.[2] Painless cervical lymphadenopathy with or without extra nodal manifestations are among the most common clinical presentations.[3] About 40% of Rosai-Dorfman disease cases have extranodal involvement with predominant head and neck region. Gastrointestinal system is among the least common sites for extranodal manifestation of RDD with only few cases reported in literature so far. Moreover involvement of liver to the extent of cirrhosis is even rarer and only one case was reported till last year.[4,5] Here we are presenting an unusual case of RDD in a middle-aged male with ascites.

Case Report

A 38-years-old gentleman was seen on 3rd February 2017 at Aga Khan University Hospital Karachi who presented complaints of weight loss and abdominal distension for 4 to 5 months. He was having low grade fever and bilateral painless neck swellings. There was no history of body rash, joint pain and pedal edema or of tuberculosis contact. His personal history, family and socioeconomic history was not significant. On examination, he looked emaciated, having distended abdomen. Vitally stable, multiple bilateral posterior cervical lymph nodes were palpable. Overlying skin was normal, non-tender, firm and rubbery in consistency 2-3 cm in size, mobile, not matted together or adherent to overlying skin or underlying structures. No axillary or inguinal lymphadenopathy pedal edema, rash or joint pains were present. There was hepatosplenomegaly with ascites. Rest of the systemic examinations was normal. Based on clinical features, initial differential diagnosis were tuberculosis, lymphoma and decompensated chronic liver disease.

Laboratory investigations revealed haemoglobin10.9 gm%, packed cell volume 34.3 %, total leukocytes count

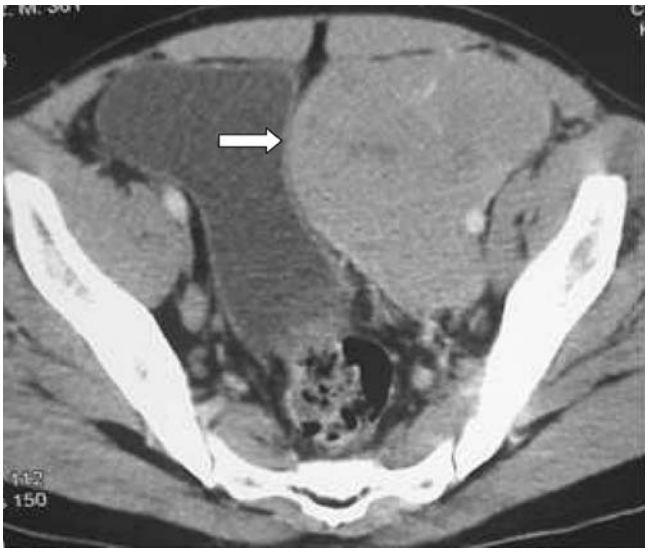

Figure-1: Arrow points at enlarged pelvic lymph node compressing urinary bladder.

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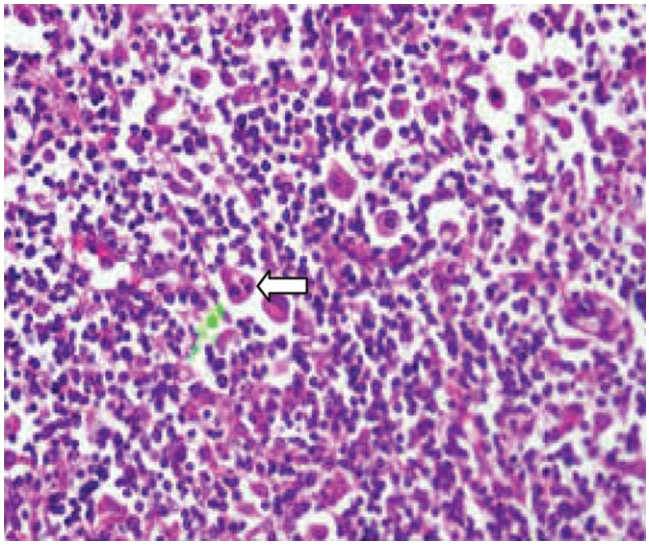

Figure-2: Arrow points at Emperipolesis/ lymphopheocytosis (20x).

2.7/mm3 with neutrophils 66.7%, lymphocytes 16.1%, eosinophils 0.4%, monocytes 16.1%, platelets counts 144,000/mm3 and erythrocytes sedimentation rate 38mm/hour. Red cells morphology was normal. Liver function test showed total bilirubin of 1.5 with direct and indirect bilirubin of 0.7 and 0.8 respectively, ALT 111, GGT 102, alkaline phosphate 141, AST 156. Serum LDH 938, Serum Albumin 2.8 and autoimmune profile negative. Routine urine examination, urine culture and blood culture were negative. Chest X-Ray was normal. Hepatitis B,C and HIV serology was negative. Abdominal imaging revealed hepatosplenomegaly, intra abdominal lymphadenopathy and ascites. Cervical lymph node biopsy showed predominantly necrosis with occasional small well-formed granulomas and palisading histiocytes. He was started on anti-tuberculosis drugs which he did not responded to and his condition continued to worsen with the development of massive ascites. Ascitic fluid analysis revealed exudative ascites, gram stain and culture negative. AFB smear and culture came negative. Cytology was negative for malignant cells.

He was scheduled for a diagnostic laproscopy and intra abdominal lymph node biopsy. His intra abdominal lymph node biopsy revealed effacement of lymph node architecture by many histiocytes mixed with eosinophils, plasma cells and vague collections of epitheloid cells. Immunohistochemistry was negative for lymphoma but CD68 and S100 protein positive. He was diagnosed as having Rosai-Dorfmann's disease, and started on oral prednisolone and referred to oncologist for further management. Informed consent was taken for this rare case to be published as a case report.

Discussion

Rosai-Dorfman disease, or sinus histiocytosis, with massive lymphadenopathy is a rare benign histiocytosis commonly affecting children and young adults, slightly more common in males — 58% for males versus 42% for females. Initially described by Destombes in 1965, this disease was highlighted as Sinus histiocytosis with massive lymphadenopathy by Rosai and Dorfman in 1969.[6,7] It is a self-limiting histiocytic proliferative disease of idiopathic origin, which is usually benign and occurs in the first two decades of life. It is assumed to be an active process or infection secondary to a virus (HHV-6) or an undefined immunological defect initiated by some other organism. Patients may present with systemic symptoms including fever, night sweats, malaise, and weight loss which may be related to enhanced production of monokines by the activated histiocytes.[1] RDD mostly presents with massive non-tender cervical lymphadenopathy. Extranodal involvement is common and is found mainly in the head and neck regions. However, it may also involve the skin, bone, kidneys, testis ,orbits, eyelids, upper respiratory tract, peritoneum, salivary glands, and central nervous system.[3] GI tract is rarely involved by RDD and represents less than 1% of extranodal presentations. Gastrointestinal RDD commonly affect ileocecal area and distal colon in middle-aged females with an over whelming majority of the cases being located beyond the pylorus.[1] Involvement of liver was previously reported in literature but RDD with liver cirrhosis was reported recently in previous year.[5]

Histopathologically, diffuse histiocytosis, homogeneous chromatin and a single small nucleolus are characteristic features of both nodal and extra nodal RDD, along with emperipolesis, which consists of the passage of intact, often multiple lymphocytes through the cell within intracytoplasmic vesicles. On the other hand, presence of prominent eosinophils or necrosis are against the diagnosis. RDD can be differentiated from Erdheim-Chester diseaseand Langerhans cell histiocytosis on the basis of lesion alhistiocytes expressing both CD68 and S-100 protein while negative for CD1a.[8]

Treatment depends upon the individual patient and is planned after determining the extent of the disease although no ideal treatment is available for all patients. It is believed that most of the patients without massive lymph nodes enlargement or organ dysfunction have spontaneous resolution of symptoms without treatment. In case of organ dysfunction, documented fever of 38 degree celsius or more without evidence of

infection or there is a sudden lymph nodes enlargement, steroids treatment should be started.[9] Rosai-Dorfman disease involving the gastrointestinal system does not spontaneously resolve.[4] Surgical interventions and/or chemotherapy can be used in patients with persistent disease or cases where organ function is threatened, such as respiratory distress or kidney failure. Radiotherapy has also been tried in some cases. Overall management goal is to use as little treatment as possible to control the disease and preserve quality of life.

Conclusion

Rosai-Dorfman Disease is a rare benign disorder which clinically presents with bilateral cervical lymphadenopathy. Extra-nodal manifestation is common but gastrointestinal system involvement is rare. The purpose of this case report is to highlight the importance of its presentation mimicking lymphoma and some endemic infectious disease like tuberculosis in developing countries such as Pakistan.

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