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Rosai-Dorfman Disease in a 23-Year-Old Patient

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Introduction

Rosai-Dorfman Disease (RDD) also known as Sinus Histiocytosis With Massive Lymphadenopathy (SHML) is a rare and benign non-Langerhans histiocytosis. First described in 1965 by Destombes [1] then in 1969 by Rosai and Dorfman, this disease is characterized by benign histiocytic proliferation with frequent lymphophagocytosis (emperipolesis) [2]. All ages are affected but most of the patients present before age 20, with a slight male predominance [3]. Rosai-Dorfman disease usually presents as bilateral, massive and painless cervical lymphadenopathy [2]. Extra-nodal involvement is found in 30-40% of cases and is more common in the head and neck region [3]. The disease is often benign and self limiting requiring only observation. In some cases, the condition can be severe. Several treatments have been used with varying efficiency [4]. We report a case of RDD in a male Tunisian patient treated and followed up in our department.

Case Report

A 23-year-old male patient, with no particular medical history, was hospitalized in our department for exploration of a biological inflammatory syndrome associated with cervical lymphadenopathy. He had no travel history and he never had transfusions or contact with animals. His family history was also unremarkable. On physical examination, the patient was a febrile and in a good general condition. His weight was of 50 Kg. He had a pulse rate of 90/min, blood pressure of 120/80 mmHg, respiratory rate of 18/min, and moderate pallor. We noted the presence of a left upper eyelid mass with incomplete mechanical ptosis, three juxtaposed left supraclavicular lymphadenopathy measuring 2 cm each one and bilateral infracentimetric right jugulo-carotid lymphadenopathy. All the adenopathies were firm, mobile, without inflammatory signs in sight. Cardiac and respiratory examinations were normal. His abdomen was soft and there was no hepatomegaly or splenomegaly. Musculoskeletal and neurological examinations were also normal. At the time of admission, hematological investigations showed hypochromic microcytic aregenerative anemia with hemoglobin of 10.2 g/dl, a total leukocyte count of 7940 cells/mm^3 (with differential count of 6140 cells/mm^3 neutrophils, 1380 cells/mm^3 lymphocytes, 70 cells/mm^3 (eosinophils), platelets of 451×10^3/mm^3, erythrocyte sedimentation rate of 120 mm/h and C-reactive protein level of 161 mg/l. Metabolic parameters were as follows: blood urea 3.34 mg/l, serum creatinine 57 umol/l, sodium 137 mmol/l, and potassium 4.2 mmol/. Liver function tests were normal: total bilirubin 5.56 umol/l, direct bilirubin 0.5 umol/dl, SGOT 18UI/l, SGPT 10 UI/l, LDH 249UI/l alkaline phosphatase 265 UI/l. Serum protein electrophoresis revealed total protein level of 66 g/l, with hypoalbuminia (22.5 g/l), hyper alpha2-globulenemia (10.2 g/l) and slight polyclonal hyper gammaglobulinemia (15.6 g/l). Serum calcium, magnesium, and phosphorus were normal. The tuberculin skin test and the direct visualisation of acid fast bacilli in gastric aspirate and urine were negative. HIV ELISA, serological viral tests (EBV, CMV, HBV, HCV), serological tests for brucellosis and the Widal test were all negative. ANA test came out positif at 1:320. A neck-chest-abdomen CT scan showed multiple bilateral cervical lymphadenopathies and homogenous hepatosplenomegaly. Endoscopic examination with biopsy of the nasopharynx showed chronic and acute inflammation of the nasopharyngeal mucosa. The left supraclavicular lymph node biopsy showed on microscopic examination a dilatation of the sinuses and lymphophagocytosis (emperipolesis) (Figure 1). The predominant cells in the sinuses were histiocytes which were positive with S-100 protein and negative for CD1a by immunohistochemistry. This histopathologic aspect was characteristic of the Rosai Dorfman disease. A CT scan of facial bones revealed the left eyelid mass that extends into the left orbit and eliminated any sinusal involvement (Figure 2).

The patient was treated with high dose prednisone (50 mg/j). One month later, a marked reduction of the cervical lymph nodes size and a regression of the systemic inflammation (erythrocyte sedimentation rate: 26 mm/h1 and C-reactive protein: 30 mg/L) were observed. Slight regression of the orbital mass was observed and surgical treatment was proposed but the patient refused it. On three month follow up the patient was well with no abnormalities on inflammatory parameters. An additional regression of the orbital mass was noted. No lymphadenopathy was found on cervical ultrasound. The orbital mass has not increased after one year follow up. Although the absence of histological confirmation we considered it as an orbital involvement of the RDD.

Discussion

Rosai Dorfman disease, otherwise known as Sinus Histiocytosis with Massive Lymphadenopathy (SHML), is a rare histiocytic-phagocytic proliferative disorder [1,2]. This disease displays a predilection for males (2:1 male-to-female ratio); usually in the first and second decades of life as it was the case of our patient. Clinically, RDD has various features and follows a chronic and indolent course [3]. Massive painless

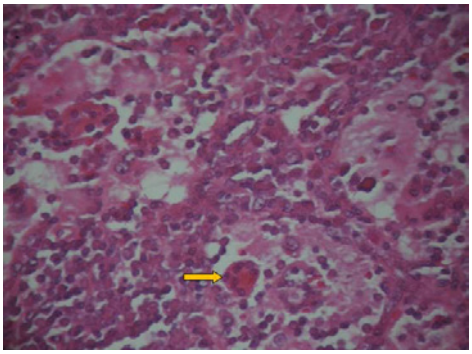

Figure 1: Lymph node biopsy showing a mixed cell population, predominantly mature histiocytes with evidence of emperipolesis (arrow).

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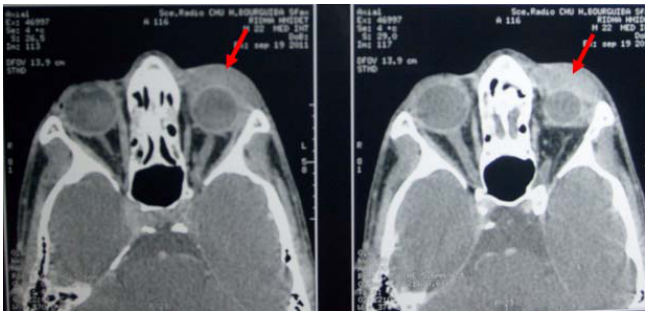

Figure 2: The CT scan of facial bones showing the left eyelid mass extending into the left orbit (arrows).

cervical lymphadenopathy is the predominant clinical manifestation of the disease (87% of cases). At early stage of the disease, lymph nodes are bilateral, mobile, and small. With disease progression, they become adherent, forming a voluminous nodular cervical mass [3]. Other lymph node groups can be affected (axillary group: 24%, inguinal group 26%, and médiastinal group 14.5%) with or without cervical involvement [5]. In our patient, bilateral cervical and sus-clavicular lymph nodes were involved without the participation of others lymph node groups. Fever is observed in 30% of cases [3]. It was absent in our case. Up to 43% of the patients have extranodal disease mainly affecting the head and neck area and exceptionally occurring without lymphadenopathy [6,7]. The most common sites of extranodal involvement, in the decreasing order of frequency, are skin (27.5%), nasal and paranasal cavities (27%), soft tissues (22%), eyelids and orbit (20%), bone (18.4%) and central nervous system (5%) [6-8].The gastrointestinal system is exceptionally affected. Hepato-splenomegaly, noted in our case, is uncommon unlike in other histiocytosis [3]. In a series of 11 patients with digestive manifestations of RDD, hepatic involvement was found in 5 cases and was associated with widely disseminated disease in the form of multiple bone and skin lesions [9]. Orbital involvement is the most common of ophthalmic RDD manifestations and is usually associated with lymphadenopathy. In a series of 113 cases of RDD, 13 patients had ophthalmic infiltrates. Eleven of the 13 had infiltrates in the orbital soft tissues, and five of these patients also had eyelid involvement [10]. Our patient had orbital and eyelid involvement without ocular compression or functional impact. Clinical laboratory findings include hematological abnormalities such as normocytic or microcytic anemia (66%), leucocytosis (59%), neutrophillia (68.5%), increased Erthrocyte Sedimentation Rate (ESR) (88.5%) and hypergammaglobulinemia (90%) [3]. Immune disorders occur in approximately 13% of patients with Rosai-Dorfman disease [3]. Antierythrocyte autoantibodies are the most common findings [10,11]. Rarely, patients exhibit rheumatoid factor, antinuclear antibodies and reversal of the CD4/CD8 ratio among peripheral lymphocytes [10,11]. Our patient showed microcytic anemia, neutrophilia, elevated ESR, hypergammaglobulinemia and positive ANA.

The diagnosis of RDD is based on histopathology [12]. Nodal sinuses are dilated and massively infiltrated by numerous large histiocytes, lymphocytes and plasma cells. Emperipolesis, characterized by the presence within the abundant histiocytes cytoplasm of viable inflammatory cells such as lymphocytes, is a suggestive but nonspecific feature of RDD. In immunohistochemical studies, the histiocytic cells are positive for S100 protein, CD14, CD68 (macrophage markers), CD11c (early dendritic cell marker), but negative for CD1a (positive on Langerhans-type of dendritic cells) [12]. The differential diagnosis includes many histiocytic disorders, such as Hodgkin's disease, malignant histiocytosis, and lymphomas as well as Langerhans cell histiocytosis. RDD, when it presents with extranodal diseases, may be misdiagnosed as a malignant disease or metastatic lymphadenopathy [12,13]. The presence of emperipolesis, absence of cellular atypia, immunohistochemical profile, and associated clinical features distinguish RDD from other simulating disorders.

Most often, RDD takes a mild course requiring only observation [4,5,14]. However, patients having extensive or progressive presentation or cosmetic abnormalities need treatment. Management options include systemic corticosteroids, chemotherapy (vinca alkaloid, alkylating agent, methotrexate and 6-mercaptopurine) or radiotherapy [4,14,15]. Good response to corticosteroid treatment was reported in many cases [4,10,14]. Surgical options are reserved for cosmetic abnormalities and or compressive symptoms like airway obstruction, neurologic or ocular compression [4,10,14]. In our patient, cervical lymphadenopathy and inflammation responded favorably to oral prednisone. Regression of the orbital mass was observed. In the absence of the histological confirmation, we considered the orbital mass as a manifestation of RDD on the basis of the response to corticosteroid treatment and the absence of recurrence one year later.

Conclusion

We report an additional case of RDD presenting with the typical manifestations of cervical lymphadenopathy associated to hepatosplenomegaly, orbital involvement and positive ANA. The diagnosis of RDD is seldom considered, mainly because of the rarity of the disease. Lymph nodes biopsy is necessary in order to differentiate this rare benign disease from malignant or inflammatory process. Treatment is not necessary in most instances. Some patients, therefore, may require corticosteroids, surgery, radiotherapy or chemotherapy because of extra-nodal spread, vital organ involvement and/or immunologic manifestations which are correlated with morbidity and mortality.

Conflict of Interests

The authors declare that they have no conflicts of interests here.

Authors' Contributions

All authors contributed to the elaboration of this paper. The authors have read and agreed upon its content and upon the fact that readily reproducible materials described in the paper will be freely available to any scientist wishing to use them for noncommercial purposes.

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