CASE REPORT

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A Case of Sarcoidosis with Atypical Presentation

Atipik Prezentasyon Gösteren Sarkoidoz Olgusu

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ÖZ

Sarkoidoz, nonkazeifiye granülomlarla karakterize, etiyolojisi bilinmeyen multisistemik bir hastalıktır. Olguların %90'ında akciğer bulguları mevcut olmakla birlikte, ekstrapulmoner sarkoidoz olguları oldukça nadirdir ve atipik klinik seyir nedeniyle tanı koymak genellikle zordur. Burada klinik takibinde Makrofaj aktivasyonu sendromu ve lenfoma ekarte edildikten sonra tanısı konulan bir ekstrapulmoner sarkoidoz olgusunu sunmayı amaçladık.

Anahtar Kelimeler: lenfoma; makrofaj aktivasyonu sendromu; sarkoidoz

ABSTRACT

Sarcoidosis is a multisystemic disease characterized by non-caseating granulomas of unknown etiology. While pulmonary manifestations are present in 90% of cases, extrapulmonary sarcoidosis cases are rare and diagnosis is often challenging due to atypical clinical course. Here, we aim to present a case of extrapulmonary sarcoidosis diagnosed after excluding Macrophage Activation Syndrome and lymphoma during clinical follow-up.

Keywords: lymphoma; macrophage activation syndrome; sarcoidosis

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INTRODUCTION

Sarcoidosis is a multisystemic granulomatous disease of unknown cause. Lung involvement is the most common presentation, but occasionally, other organs can be affected without lung involvement. Diagnosing cases without lung involvement can be particularly challenging due to the disease's multisystemic nature, which mimics various other conditions, and the absence of clear diagnostic criteria. Consequently, patients often undergo numerous advanced tests, leading to delays in diagnosis and increased medical costs.[1]

In this case report, our aim is to emphasize that sarcoidosis should be considered in the differential diagnosis of patients presenting with nonspecific symptoms such as prolonged fever, weight loss, and skin rash.

CASE REPORT

A 35-year-old female patient with no medical history was admitted to the city hospital with skin rash, involuntary weight loss and fever attacks going on for about 3 months. Since the examinations performed in the hospital revealed an increase in acute phase reactants, the patient was followed up with empiric antibiotic therapy (Moxifloxacin 400 mg tb 1x1, 7 days). Due to persistent clinical complaints, she was referred to our clinic for further evaluation and treatment.

In the detailed anamnesis, the patient was married, a mother of one child, lived in a rural area, and had no family history other than hypertension in her father. In the system query, she lost approximately 10 kilos in the last 4 months, had fever and rash attacks during this period but the fever and rash attacks were not related. On physical examination, body temperature: 37.8 ^{0}C, pulse: 98/min, blood pressure: 116/73 mmHg, respiration rate: 16/min, traube's space was closed. There were widespread, patchy, salmon-coloured, macular and dandruff looking lesions on the skin of back, abdomen and legs (Figure 1).

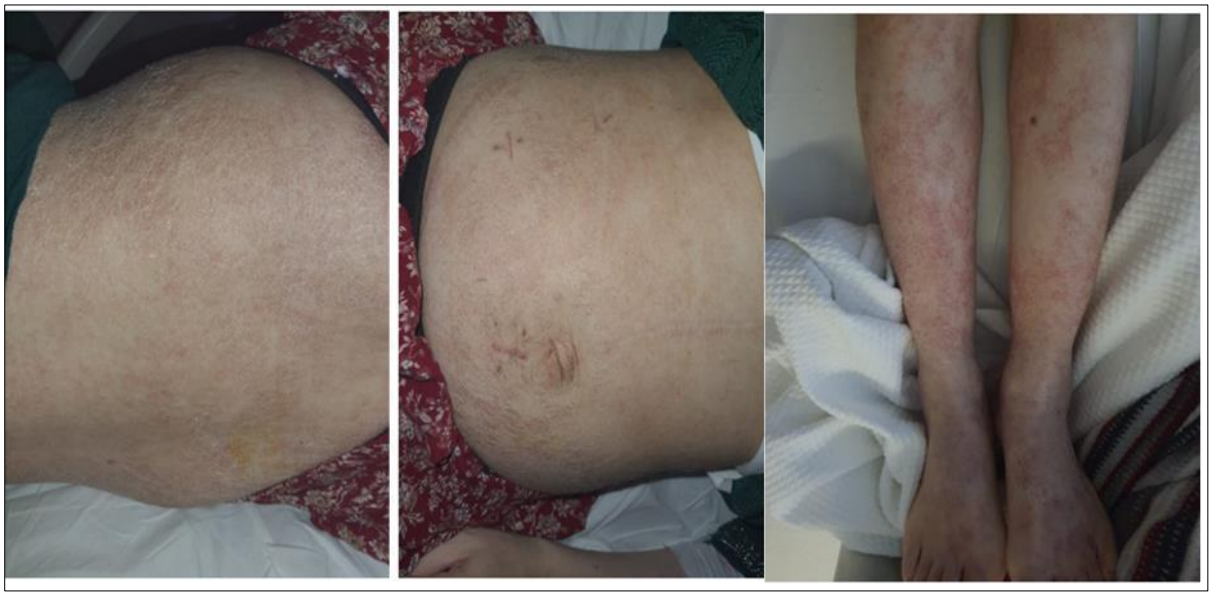

Figure 1. Skin lesion.

In routine laboratory examinations, it was observed that WBC: 2800 /mm3, NEU: 1800/mm3, LYM: 690/mm3, HGB: 9.7 g/dl, PLT: 106.000/mm3, HTC: 30, MCV: 74, CRP: 60 mg/L, ESR: 14 mm/hour, kidney function tests, liver function tests and ferritin were within normal ranges. Electrocardiography showed sinus tachycardia. In the posterior-anterior chest radiograph, the hilus appeared normal and there was no sign of active infiltration or fluid (Figure 2). Abdominal ultrasonography showed that the length of the right lobe of the liver was craniocaudal 17.5 cm and the long axis of the spleen was 21 cm.

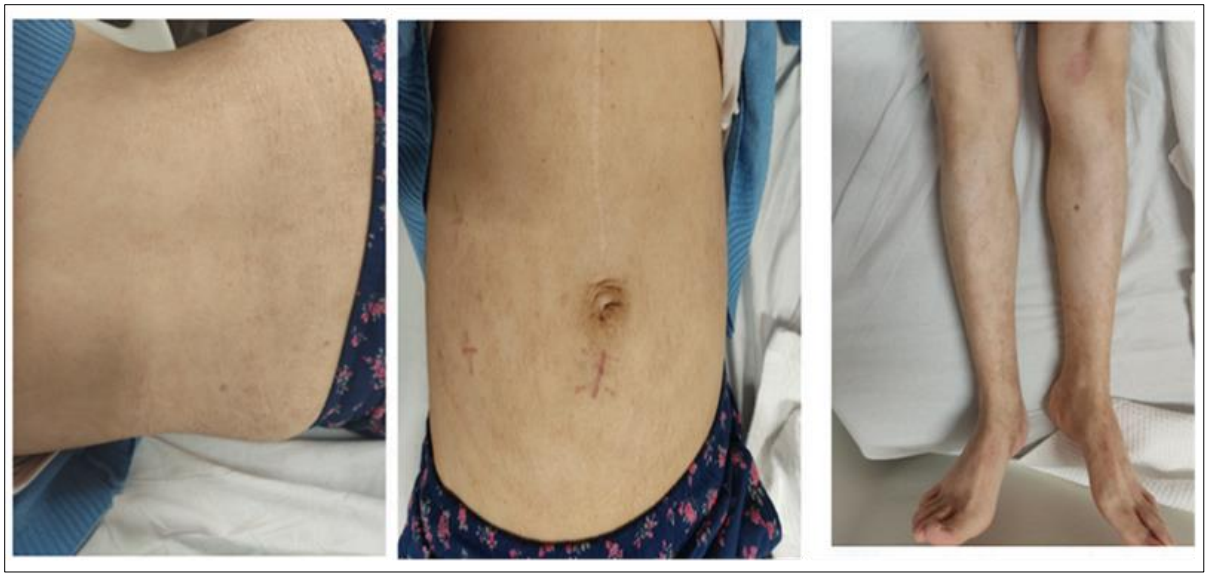

Figure 2. Posterior-anterior chest radiograph.

Since considered lymphoma, rheumatological disease and systemic infective conditions, rheumatological panel, Interferon Gamma Release Test (IGRA), Franciella tube agglutination test (Tularemia), rose bengal slide agglutination test (Brucella), VDRL test (Syphilis), Grubel-Widal agglutination test (Salmonella), TORCH panel, hepatitis panel was performed and all test's results were negative. The PPD test interpreted as anergic. No growth was detected in the blood and urine cultures. Superficial ultrasound performed for lymphoma revealed suspiciously reactive lymphadenopathies in the left cervical, right and left submandibular and left and right ingunal regions, the largest of which did not exceed 18 mm. Therefore, PET-CT was planned for the patient. In PET-CT, the spleen had a heterogeneous appearance and increased uptake (Suvmax: 5.7). No involvement was observed in other organs.

It was decided to perform a bone marrow biopsy to rule out lymphoma and macrophage activation syndrome (MAS) for the patient because of pancytopenia and fever continued during follow-up. Since the biopsy result showed "normocellular bone marrow with a slight increase in megakaryocytes" it was decided to biopsy the patient's rash. The skin biopsy showed "perivascular chronic cells in the superficial dermis, inflammatory cell infiltration, and granuloma structures without necrosis in several areas of the dermis." The patient was re-evaluated for granulomatous diseases and the ACE level was sent for sarcoidosis. The patient's serum ACE level was 299.1 so that 40 mg/day prednol treatment was started with the diagnosis of extrapulmonary sarcoidosis. The skin rash was observed to regress after treatment (Figure 3).

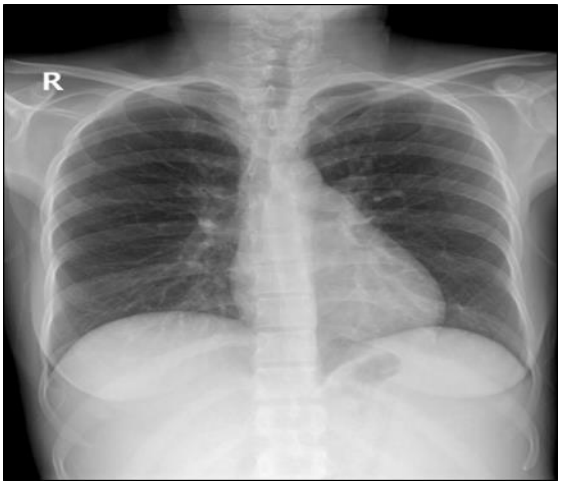

Figure 3. Skin lesions after treatment.

However, pancytopenia condition persisted and the patient was evaluated by the council consisting of hematology, rheumatology and chest diseases. It was decided to perform splenectomy to rule out isolated spleen lymphoma and treat pancytopenia. Biopsy result was compatible with splenic sarcoidosis. In addition to steroid treatment, plaquenil and azathioprine were started. It was seen that pancytopenia condition of the patient improved in several days and no fever was observed during follow-up. She was discharged with full recovery after oral medication was adjusted and internal medicine outpatient clinic check-up was recommended.

Informed consent was obtained from the patient.

DISCUSSION

Sarcoidosis is a multisystemic granulomatous disease of unknown origin. Its pathophysiology involves an unknown antigen triggering a granulomatous reaction through stimulation of stem cells. While lung involvement occurs in 95% of cases, extrapulmonary sarcoidosis is observed in about 2% of cases. Due to the lack of clear diagnostic criteria, particularly in extrapulmonary cases, it can mimic various other diseases, leading to diagnostic challenges.[1-3]

Skin is the second most commonly affected organ after the lungs in sarcoidosis. In many cases, skin involvement does not cause symptoms beyond cosmetic appearance. However, studies indicate that systemic involvement can develop in approximately one-third of patients with cutaneous sarcoidosis.[3,4]

Skin involvement in sarcoidosis can manifest in specific and nonspecific lesions. Nonspecific lesions such as erythema nodosum, psoriatic-like lesions, and verrucous lesions typically do not show granulomas on biopsy. In contrast, specific lesions like maculopapular rashes, subcutaneous nodules, and Lupus pernio exhibit characteristic noncaseating granulomas upon biopsy.Traditionally, the treatment approach for skin lesions in sarcoidosis involves the use of topical and systemic steroids.[4,5] In our case, maculopapular rash, one of the specific skin findings of sarcoidosis, was found on the patient's back, abdomen and legs. Granulomas without necrosis were detected in the skin biopsy and it was observed that the skin findings regressed dramatically after prednol treatment.

Splenic involvement in sarcoidosis occurs 3% of the patients and is usually asymptomatic.[6] Rarely, it may cause massive splenomegaly and pancytopenia and so that symptoms of thrombocytopenia, anemia and leukopenia may be observed in patients as a result of hypersplenism.[7] However, studies have observed that extrathoracic symptoms (skin lesions, joint symptoms, hepatomegaly and peripheral adenopathy) are more common in sarcoidosis patients with significant splenomegaly. While splenic involvement in sarcoidosis largely responds dramatically to treatment, splenectomy may be required intreatment-resistant cases.[8] In our case, the patient had splenomegaly and pancytopenia but it was observed that the pancytopenia persisted despite steroid treatment. Hence, the patient underwent splenectomy for the treatment of corticosteroid-resistant pancytopenia and it was observed that the pancytopenia improved after splenectomy.

Since sarcoidosis is a disease that can affect all organs and systems, atypical features of sarcoidosis can be present in a wide range. In such patients nonspecific findings may be observed in biochemical and radiological examinations.[9] For this reason, patients are exposed to many unnecessary examinations and have delayed diagnosis. In our case, the patient was diagnosed after several rheumatological, hematological and infectious diseases were excluded by many examinations.

As a result, it is important to keep in mind the atypical clinical presentations of sarcoidosis in patients with multicytemic findings and without a clear diagnosis, in order to make a rapid diagnosis and to protect patients from unnecessary examinations.

Conflict of Interests

The authors declare that there is not any conflict of interest regarding the publication of this manuscript.

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Ethics Committee Permission

Informed consent was obtained from the patient.

Authors' Contributions

Concept/Design: MVC, DA. Data Collection and Processing: DA, ACK. Data analysis and interpretation: MVC, SK. Literature Search: ACK, DA. Drafting manuscript: MVC, ACK. Criticalrevision of the manuscript: SK, MVC. Supervision: SK.

REFERENCES

1. Judson MA. The Clinical Features of Sarcoidosis: A Comprehensive Review. Clin Rev Allergy Immunol. 2015;49(1):63-78.

2. Sharma OP. Sarcoidosis: clinical, laboratory, and immunologic aspects. Semin Roentgenol. 1985;20(4): 340-355.
3. Rao DA, Dellaripa PF. Extrapulmonary manifestations of sarcoidosis. Rheum Dis Clin North Am. 2013;39(2):277-297.
4. Marchell RM, Judson MA. Cutaneous sarcoidosis. Semin Respir Crit Care Med. 2010;31(4):442-451.
5. Wanat KA, Rosenbach M. Cutaneous Sarcoidosis. Clin Chest Med. 2015;36(4):685-702.
6. Selroos O. Sarcoidosis of the spleen. Acta Med Scand. 1976;200(1-6):337-340.
7. Kataria YP, Whitcomb ME. Splenomegaly in sarcoidosis. Arch Intern Med. 1980;140(1):35-37.
8. Sharma OP, Vucinic V, James DG. Splenectomy in sarcoidosis: indications, complications, and long-term follow-up. Sarcoidosis Vasc Diffuse Lung Dis. 2002;19(1):66-70.
9. Modi T, Maniam G, Quirch M, et al. Extrapulmonary sarcoidosis in an atypical patient demographic. Proc (Bayl Univ Med Cent). 2020;34(1):151-152.