Adult-onset Still's disease and dermatomyositis, is there a relation?

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Adult-onset Still's disease (AOSD) is a rare systemic inflammatory disorder of unknown etiology. We report a 22-year-old female patient, admitted to our department with fever, rash, large joint pains, and mild proximal weakness of both upper and lower limbs. With further evaluation of the patient and exclusion of infections, hematological malignancies, and connective tissue disorders, AOSD diagnosis was established with strikingly elevated serum ferritin level. This patient is the daughter of a patient who was diagnosed as having dermatomyositis in our department 2 years earlier. The mother was re-evaluated, and AOSD could not be excluded through the normal serum ferritin level.

Keywords:
adult-onset Still's disease, dermatomyositis, myopathy, neurological manifestations

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Introduction

Adult-onset Still's disease (AOSD) is a rare systemic inflammatory disorder of unknown etiology that is frequently underdiagnosed and typically characterized by a clinical trial of daily spiking high fever, evanescent rash (salmon colored), and arthritis. A biological triad of hyperferritinemia, hyperleukocytosis with neutrophilia, and abnormal liver function test result also characterizes the disease.

There is no specific test or a combination of tests that establishes the diagnosis of AOSD; however, the clinical presentation together with elevated serum ferritin is highly suggestive of AOSD [1].

The incidence of AOSD is estimated to be 0.16 per 100 000 persons, with a bimodal peak at ages 15–25 and 36–46 years (with three-quarters of the patients reporting disease onset between 16 and 35 years of age) and equal distribution between males and females [2].

Although the exact etiology is still unknown, some observations supported a genetic predisposition and environmental factors that trigger the immune system (immune mediated disease); a range of infectious agents may trigger the disease in a genetically predisposed host [1].

Case report

A 22-year-old female patient presented with multiple large joint pains and proximal weakness of both upper and lower limbs. She had spiking fever as well, especially in the evening reaching 39.5°C, associated with severe sore throat 2 weeks before the onset. The onset of fever was also associated with an evanescent, pruritic erythematous linear rash mainly over the trunk and extremities. Her medical history was unremarkable, and her mother was a known case of dermatomyositis.

Examination revealed fever of 39.0°C. There was lymphadenopathy but no splenomegaly. She had acute synovitis of wrists, elbows, ankles, and knees. There was a residual skin rash seen over the trunk and extremities.

Neurological examination finding was free of any abnormalities apart from muscle tenderness and mild proximal weakness in both upper and lower limbs grade 4, with brisk deep tendon reflexes.

Hematological investigations showed leukocytosis of 8×10^9/l with neutrophilia. Erythrocyte sedimentation rate was high 80/115 mm/h. There were markedly elevated levels of serum ferritin (1616.6 ng/ml). The findings of antinuclear antibody, antidouble-strand DNA, and rheumatoid factor were all negative. Renal and liver function findings as well as coagulation profiles were normal. Blood and urine cultures revealed no evidence of bacterial, fungal, or viral infection.

Electromyogram showed evidence of myopathy.

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Based on the clinical features and laboratory results, she was diagnosed as having AOSD according to the Yamaguchi criteria [3].

The patient was treated with corticosteroids, initially intravenously and then orally, with improvement in her symptoms.

The mother of our patient who was diagnosed as having dermatomyositis 2 years ago, with typical skin eruption and inflammatory myopathy, was asked to visit us to undergo laboratory tests, serum ferritin together with erythrocyte sedimentation rate, antinuclear antibody, and rheumatoid factor. The results of which came out negative. We revised the literature aiming at finding relation between AOSD and dermatomyositis, and there were some reports of AOSD presented with dermatomyositis like skin rash [4].

Having low serum ferritin level does not exclude the diagnosis of the mother as having AOSD as ferritin level is lower in remission, and in ~10% of the patients with AOSD, the ferritin levels are within reference range [5].

Although it is known that AOSD is a sporadic noninherited disease, we report a patient and her mother who had myopathic syndromes with skin rash; for the patient, the most likely diagnosis is AOSD.

Discussion

AOSD is a rare inflammatory disorder that has been recently classified as a polygenic autoinflammatory disorder. AOSD can be divided into two phenotypes based on cytokine profile, 'systemic' and an 'articular' pattern. Interleukin-1 (IL-1) family (namely, IL-1β and IL-18) plays a major role in the pathophysiology of the systemic pattern of the disease. Drugs that antagonize IL-1β, like anakinra, have been used successfully in the treatment of resistant cases of AOSD [6]. Revising the literature revealed that IL-1β expression is one of the most consistent cytokine patterns in the muscle tissue of patients with dermatomyositis. Treatment with anakinra had a beneficial effect in a series of patients with refractory DM [7]. So it is clear now that both diseases share common pathophysiological aspects.

AOSD has different neurological presentations. The nervous system was affected in 7–12% of cases, mainly as cranial nerve palsies, pyramidal and extrapyramidal affection, aseptic meningoencephalitis, demyelinating encephalopathy, seizures, myopathy, and Miller-Fisher syndrome have been also described [8–10].

The diagnosis is often difficult as the condition is similar to systemic infections, vasculitis, or even some malignant condition; to establish the diagnosis of AOSD, these condition should be excluded, and elevated levels of serum ferritin may help [3]. Our patient presented with almost all the major and minor criteria together with abnormally high serum ferritin level, and her mother has been diagnosed and treated as diabetes mellitus.

Conclusion

AOSD can present for the first time at the neurology clinic and should be included in the differential diagnosis of young patients presenting with high fever with muscle pain and weakness associated with arthritis. The diagnosis of AOSD is done by exclusion after investigations for other similar conditions, for example, infections, granulomatous disorders, connective tissue disorders, and malignancies like lymphoma.

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Conflicts of interest

There are no conflict of interests.

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