









Case Report

Juvenile Systemic Sclerosis Complicated by Interstitial Lung Disease and Myositis: A Case Report

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Abstract

Systemic sclerosis (SSc) is a rare connective tissue disorder characterized by fibrosis, vascular alterations, and organ dysfunction. Juvenile systemic sclerosis (jSSc), an infrequent form of the disease, primarily affects children, with an incidence of 0.27 to 1 per million. This report details the case of a 10-year-old male patient diagnosed with jSSc complicated by interstitial lung disease (ILD) and myositis. The patient initially presented with characteristic signs of skin thickening and Raynaud's phenomenon, which later advanced to the emergence of ILD and myositis. The case emphasizes the importance of vigilant screening for ILD in jSSc patients, given the severity and associated increased mortality of the complication, and improves our understanding of the underlying clinical features of this disease. It also calls attention to the challenges of managing the disease effectively and necessitates further research to improve treatment outcomes for such patients.

Keywords: Interstitial lung disease, Juvenile systemic sclerosis, Mycophenolate mofetil, Myositis, Systemic sclerosis.

التصلب الجهازى الشبابى المعقد بمرض الرئة الخلالى والتهاب العضلات: تقرير حالة

الخلاصة

التصلب الجهازى (SSc) هو اضطراب نادر فى النسيج الضام يتميز بالتليف والتغيرات الوعائية واختلال وظائف الأعضاء. يؤثر التصلب الجهازى الشبابى (jSSc)، وهو شكل نادر من المرض، فى المقام الأول على الأطفال، مع حدوث 0.27 إلى 1 لكل مليون. يفصل هذا التقرير حالة مريض يبلغ من العمر 10 سنوات تم تشخيصه بمرض jSSc المعقد بمرض الرئة الخلالى (ILD) والتهاب العضلات. أظهر المريض فى البداية علامات مميزة لسماكة الجلد وظاهرة رينود، والتي تطورت لاحقا إلى ظهور ILD والتهاب العضلات. تؤكد الحالة على أهمية الفحص اليقظ لـ ILD فى مرضى jSSc، بالنظر إلى شدة المضاعفات وما يرتبط بها من زيادة معدل الوفيات، وبحسن فهمنا للسمات السريرية الأساسية لهذا المرض. كما يلفت الانتباه إلى تحديات إدارة المرض بشكل فعال ويتطلب المزيد من البحوث لتحسين نتائج العلاج لهؤلاء المرضى.

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INTRODUCTION

Scleroderma (systemic sclerosis) is a rare systemic autoimmune disease characterized by extensive fibrosis, vascular alterations, and a wide array of internal organ dysfunction, including the lungs, gastrointestinal tract, heart, and kidneys [1]. It is considered a connective tissue disorder of unknown etiology with chronicity and usually a progressive course [2]. The organ manifestations showed some differences as well as some similarities in their incidence between the adult and juvenile types of this disease. The incidence of ILD is similar in both the juvenile and adult subtypes, while the musculoskeletal involvement revealed some differences, such as arthritis and myositis, while increasing frequency has been observed in jSSc [3,4]. In addition to the observed increase in morbidity and mortality associated with SSs, approximately a 10-year mortality rate of around 40% was noticed in one-fourth of those who develop progressive ILD [5]. SSs is rare in children; the estimated annual incidence is 0.27 to 1 per million children, with a predominance of three to four females to one male [6]. To the best of our knowledge, there have only been a few reports of ILD in jSSc. [7,8]. In this article, we describe a rare instance of jSSc in a male patient who also had ILD and myositis, based on the clinical manifestations associated with the disease's emergence. Additionally, it highlights the importance of screening for ILD in patients with jSSc.

Case scenario

This presentation outlines the case of a 10-year-old male patient who exhibited progressive skin tightening with subclinical ILD. The onset of the patient's symptoms may be traced back to a period of four years ago, during which there was skin tightening and itching in both hands with bluish discoloration of the fingertips (Raynaud's phenomenon) upon exposure to cold temperatures. Over time, this tightness extended to encompass the upper and lower extremities, face, and complete body. In addition to suffering from dysphagia of both solids and liquids associated with gastroesophageal reflux, as well as occasional episodes of diarrhea, cough, and polyarthralgia, there was no history of chest pain, exertional dyspnea, or passing dark urine. Family history wasn't relevant. Methotrexate at a weekly dose, a proton pump inhibitor, and amlodipine were prescribed for the patient with an insufficient response. Three months later, the patient developed difficulty standing from a sitting position and difficulty climbing stairs and was referred to the rheumatology clinic of Baghdad Teaching Hospital, where further assessment was done. Physical examination revealed a thin, pale boy with tightened skin of the face, trunk, upper and lower extremities, restricted mouth opening, nose beaking, and obvious sclerodactyly with shiny taut hypopigmented skin over proximal interphalangeal and

metacarpophalangeal joints with ischemic ulcers and scars, as shown in Figure 1 (A and B).

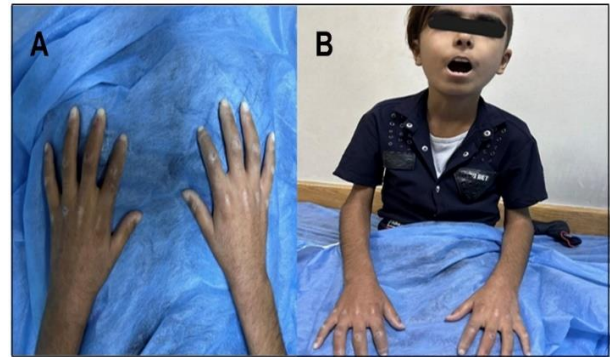


Figure 1: A) Sclerodactyly with shiny, taut hypopigmented skin over proximal interphalangeal and metacarpophalangeal joints with ischemic ulcers and scars. B) Restricted mouth opening with a beaking nose.

An assessment of the patient's musculoskeletal system revealed a flexion deformity, movement restriction at the elbows and interphalangeal joints, generalized muscle wasting, and proximal muscle weakness of grade 4 on the MRC scale, but normal power in the distal muscle groups. The chest examination was normal, apart from bilateral posterior fine-end inspiratory crepitations. The laboratory analysis revealed a decrease in hemoglobin level to 9.87 (normal: 11.5–15.5 g/dL), an increase in platelet counts up to 532 (normal: 150–400), normal WBC levels, and an erythrocyte sedimentation rate of 24 mm/h (normal: 0–20). Urinalysis, parathyroid hormone, renal, liver, and thyroid functions were within the normal range. A markedly elevated creatinine phosphokinase (CPK) level of 2737 U/L (normal, 30–200 U/L) The serology for antinuclear antibodies (ANA) and extractable nuclear antigen (ENA) panels was negative. Pulmonary function tests showed severe restrictive patterns with a forced vital capacity (FVC) of 0.62 l (reference value = 1.88 l), a forced expiratory volume (FEV 1.0) of 0.62 l (reference value = 1.45 l), and a FEV 1.0/FVC of 99.34% (reference value = 76.45%). The computed tomography (CT) scan of the chest revealed the presence of bilateral peripheral posterior lower lobe honeycombing, as shown in Figure 2 (A and B). The airways were found to be patent, with no evidence of mediastinal lymphadenopathy or pleural effusion. Additionally, no bone lesions were observed, and the echocardiogram examination yielded normal results. The electromyography findings were consistent with a chronic, non-inflammatory, non-dystrophic myopathic process affecting mainly the proximal muscles, mild to moderate in degree. A thorough evaluation of the patient's medical history, clinical examination, laboratory results, and imaging results led to the diagnosis of jSSc with ILD and myositis. The treatment plan was to start Mycophenolate Mofetil (MMF) tablets (1 g/d) and prednisolone tablets at a dose

of 1 mg/kg, with subsequent tapering with proton pump inhibitors. Two months later, his muscle power was grade 5 on the MRC scale, his CPK level dropped to 202 U/L (normally 30–200 U/L), and the patient underwent monthly follow-up.

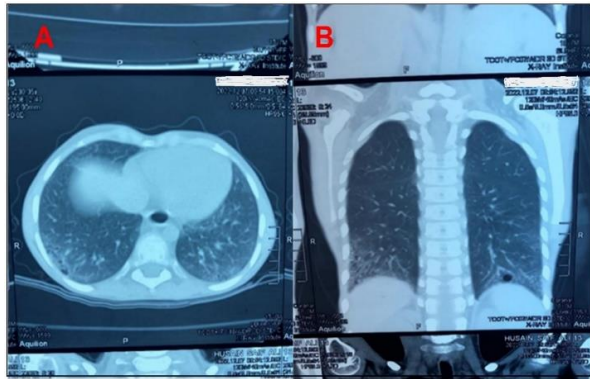


Figure 2 (A and B): A Computed Tomography (CT) scan of the chest revealed the presence of bilateral peripheral posterior lower lobe honeycombing as well as a patulous whole-length esophagus.

DISCUSSION

The most common cause of death for patients with scleroderma is pulmonary involvement—ILD in particular—followed by pulmonary arterial hypertension and cardiac involvement [9]. This patient had a sign of ILD, which was shown on a high-resolution computed tomography (HRCT) scan as fibrotic changes and honeycombing that took up more than 20% of the total lung capacity. Even though the patient did not exhibit substantial pulmonary symptoms (Figure 2A and 2B), the prevalence of ILD is frequently underestimated, and this depends on the method employed to identify fibrotic changes. A recent study on pulmonary involvement in JSSc demonstrated pulmonary involvement in the majority of patients. Additionally, patients with pulmonary involvement typically receive a diagnosis in the later stages of the disease. Notably, pulmonary fibrosis can develop silently over the course of the disease. Therefore, by the time clinical symptoms are noticeable, structural remodeling is already established and often advanced. Chronic coughing and dyspnea are the most common symptoms. In asymptomatic patients, the diagnosis of pulmonary disease in JSSc is frequently established since only a few patients present with dyspnea or a persistent cough [10]. High-resolution computed tomography, pulmonary function tests, or chest radiography can identify a high percentage of pulmonary compromise in children. Previous research has demonstrated that HRCT is the most sensitive imaging modality for lung involvement in SSc and is regarded as the gold standard for identifying specific pulmonary abnormalities [7]. Patients with myopathy frequently experience symmetrical proximal weakness, particularly in the shoulder girdle and humeral muscles,

occasionally along with noticeable atrophy. There are signs of myositis, such as higher levels of muscle enzymes and changes in the MRI of muscle groups that look like they happen in juvenile dermatomyositis. SSc muscle biopsy results are different from those of juvenile dermatomyositis in a number of ways, including a higher level of fibrosis and the presence of thickened capillaries [11]. Children who have been diagnosed with diffuse cutaneous SSc and myositis are more likely to have serious heart problems like myocardial perfusion deficits and dilated cardiomyopathy [12]. The use of rituximab for B-cell depletion is a viable option during exacerbations of progressive SSc-ILD. However, a 2019 prospective study comparing 254 patients enrolled in the EUSTAR database [13] showed that patients receiving rituximab combined with MMF had better long-term outcomes [14]. The efficacy of other immunosuppressive therapies is being studied. A randomized, controlled, double-blind, parallel-group trial comparing MMF versus oral cyclophosphamide in the treatment of scleroderma-related ILD showed significant improvement in lung function at 24 and 12 months, respectively. Furthermore, MMF was much more tolerated than cyclophosphamide, with fewer reported side effects, including leukopenia and thrombocytopenia, but they were both equally effective in improving other symptoms of skin thickness, self-reported dyspnea, and the quantitative extent of ILD in the whole lung [15]. Therefore, MMF was selected as the immunosuppressive medication for our patient in order to address ILD, myositis, and skin symptoms. This treatment plan also involves gradually reducing the dosage of steroids and scheduling monthly check-ups to monitor the patient's progress.

Conclusions

This report details a rare instance of a 10-year-old boy with jSSc who also has ILD and myositis. Early detection and treatment are vital, especially with life-threatening complications like ILD. HRCT proved essential for early ILD detection. The patient responded well to treatment with MMF. This case emphasizes the importance of ongoing research for better disease management in such rare conditions.

Conflict of interests

No conflict of interest was declared by the authors.

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Data sharing statement

Supplementary data can be shared with the corresponding author upon reasonable request.

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