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Systemic sclerosis sine scleroderma in a woman with centromere antinuclear antibodies, pulmonary and digestive involvement: Case report

Freddy Liñán Ponce
Hospital Víctor Lazarte Echegaray. Trujillo, Perú

Juan Leiva Goicochea
Hospital Víctor Lazarte Echegaray. Trujillo, Perú

José Chávez Corrales
Hospital Edgardo Rebagliati Martins. Lima, Perú

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SYSTEMIC SCLEROSIS SINE SCLERODERMA IN A WOMAN WITH CENTROMERE ANTINUCLEAR ANTIBODIES, PULMONARY AND DIGESTIVE INVOLVEMENT: CASE REPORT

ESCLEROSIS SISTÉMICA SINE ESCLERODERMIA EN UNA MUJER CON ANTICUERPOS ANTINUCLEARES CENTROMÉRICOS, AFECTACIÓN PULMONAR Y DIGESTIVA: REPORTE DE CASO

Freddy Liñán Ponce [1,a], Juan Leiva Goicochea [1,a], José Chávez Corrales [2,a]

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ABSTRACT

Introduction: Sine entities are rare in rheumatology. In progressive systemic sclerosis there is a variant of the limited cutaneous form called systemic sclerosis sine scleroderma, whose central feature is the lack of skin involvement, but visceral involvement is present. Positive anti-topoisomerase or anti-centromere antibodies confirm the diagnosis. We present the case of a 63-year-old woman with interstitial lung disease, intestinal transit involvement and Raynaud's phenomenon, with high titers of ANA with a centromeric pattern and positivity for anti-topoisomerase antibodies. When faced with a patient with Raynaud's phenomenon, visceral involvement and elevated ANA, specific antibodies should be requested for the diagnosis of systemic sclerosis in its sine variety.

Key words: Systemic sclerosis. Raynaud's disease, antinuclear antibodies. (Source: MESH-NLM)

RESUMEN

Introducción: Las entidades sine son poco comunes en reumatología. En la esclerosis sistémica progresiva, hay una variante de la forma cutánea limitada llamada esclerosis sistémica sine esclerodermia, cuya característica central es la falta de afectación cutánea, pero compromiso visceral presente. La positividad a los anticuerpos antitopoisomerasa o anticentromérico confirman el diagnóstico. El caso de este estudio es una mujer de 63 años con enfermedad intersticial pulmonar, afectación del tránsito intestinal y fenómeno de Raynaud, con ANA a títulos elevados con patrón centromérico y positividad para anticuerpos antitopoisomerasa. Ante un paciente con fenómeno de Raynaud, afectación visceral y ANA elevado, se le debe pedir anticuerpos específicos para diagnóstico de esclerosis sistémica en su variedad sine.

Palabras clave: Esclerosis sistémica, enfermedad de Raynaud, anticuerpos antinucleares (fuente: DeCS-BIREME)

[1] Hospital Víctor Lazarte Echegaray. Trujillo, Perú
[2] Hospital Edgardo Rebagliati Martins. Lima, Perú
[a] Médico asistente de Reumatología.

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INTRODUCTION

In rheumatology, there are few entities with the sine condition (Latin locution meaning "without"); they are rare diseases in which the most florid characteristic that defines the entity is absent; for example, absence of weakness in polymyositis or cutaneous sclerosis in progressive systemic sclerosis (PSS). PSS is a disease in which idiopathic fibrosis is the rule. Its hallmark is skin sclerosis, the presence of which greatly facilitates its rapid and accurate diagnosis. It is classified into limited cutaneous (SSlc), diffuse cutaneous (SSdc), prescleroderma, and systemic sclerosis sine scleroderma (SSss)[1].

Ssss is a rare entity of PSS in its limited variety (2-10% of cases) and is recognized as a variant of PSS, its diagnosis is determined by Raynaud's phenomenon (RyP), visceral involvement and specific antibodies present: Antibodies anticentromeric (AAC) or antitopoisomerase (AAT) antibodies, also called Anti-Scl 70. SSss is a rare entity, diagnosis is usually delayed, a problem that leads to a progression of complications (inexorable advance of pulmonary and digestive involvement). Therefore, in a patient with Raynaud's phenomenon, pulmonary involvement (fibrosis and/or pulmonary hypertension) and involvement of the digestive tract (gastroparesis, gastroesophageal reflux, constipation, dysphagia, heartburn, etc.), AAC or AAT should be investigated to establish the diagnosis of SSss [2]. Although it is true, in SSss, there is no skin involvement, this does not differ in the other clinical or laboratory characteristics that may exist, so the prognosis is as similar as in PSS with skin involvement. The treatment of an SSss also does not differ in the classic form of PSS, but the rule is that almost all patients with SSss begin treatment late, when the visceral fibrosis (pulmonary, digestive, renal) has already generated irreversible damage, due to the rarity of this entity[3]. The objective and justification of this report is to relate an elevated ANA with a centromeric pattern associated with pulmonary and digestive involvement with a rapid diagnostic suspicion of scleroderma sine scleroderma, a frankly rare entity.

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A 63-year-old woman, married, housewife, from the Peruvian Amazon. She has no pathological history. The patient denies smoking and alcoholism. Sporadic dry cough began, without fever or weight loss. The cough persisted and became chronic over time. Eight months after the onset of the symptoms, progressive tiredness and fatigue appeared, so she went to the hospital where a physical examination of the respiratory system revealed diffuse velcro-type crackles in the lower 2/3 of both lung fields. The chest X-ray confirmed pulmonary fibrosis, in a ground glass pattern (figure 1).

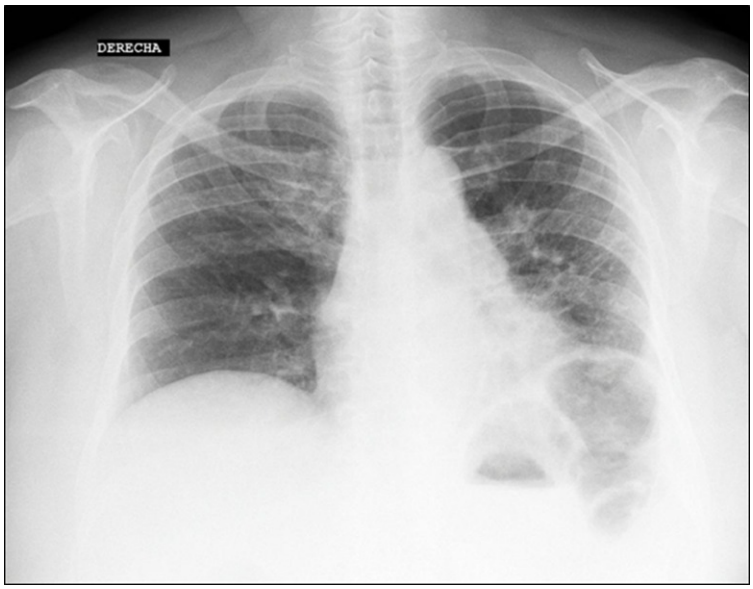

Figure 1. Chest X-ray showing a diffuse ground glass pattern in the lower 2/3 of both lung fields.

In addition, the patient begins postprandial discomfort, feeling fullness and dysphagia. There is no variation in appetite. The patient is referred to a national hospital in Lima and a study of the cause of pulmonary fibrosis and dyspepsia begins.

In the auxiliary tests, the results are normal, except for high titers of Antinuclear Antibodies (ANA) at 1/5120 with a centromeric pattern. With this result, a study of autoimmune disease was initiated. The physical examination of the abdomen was not contributory. A chest Computed Tomography (CAT) was performed (figure 2) where reticular-type interstitial thickening was reported, predominantly posterobasal subpleural .

During the hospital stay, Raynaud's phenomenon was verified repeatedly, and no other skin lesions were evident. The patient's hands also did not show distinguishable or characteristic lesions (figure 3). Contrast radiography of the digestive tract was performed (figure 4) and a delay in emptying was demonstrated at higher levels (esophagus and stomach), but no dilatations were seen. He underwent digital capillaroscopy, which revealed splinter hemorrhages in the 3rd finger of the left hand.

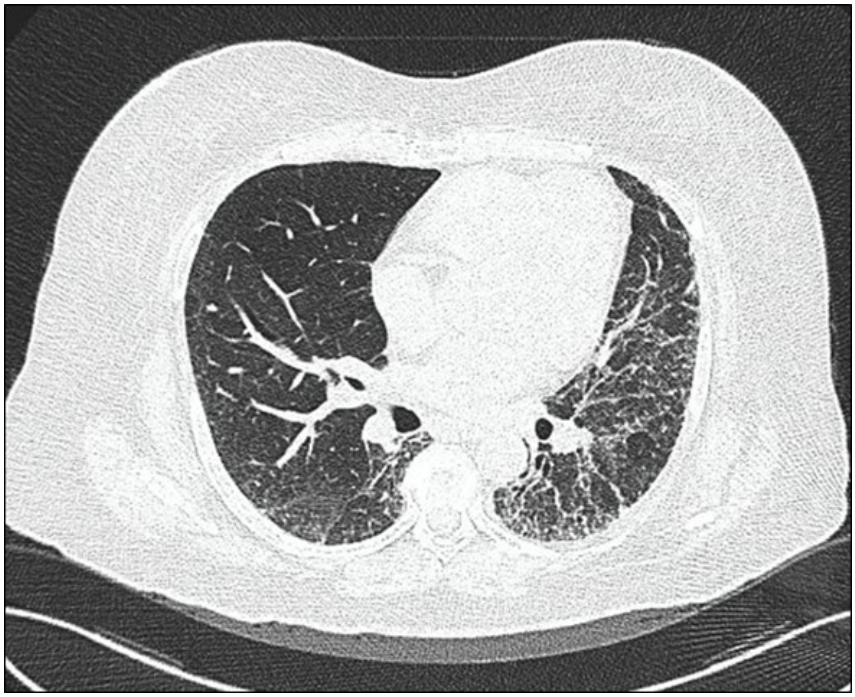

Figure 2. Progressive honeycombing to a subpleural predominance is evidenced, the reason for the tiredness and fatigue in the patient.

Given the existing triad (pulmonary fibrosis, digestive involvement and Raynaud's phenomenon), a sine entity is probably diagnosed. A study of antibodies for systemic sclerosis is requested (anti-centromeric antibodies -AAC- and anti-topoisomerase -AAT-). The result showed elevated levels of AAT. It is concluded in a Systemic Sclerosis sine scleroderma (SSss). Treatment was started with azathioprine 100 mg daily, management that causes digestive intolerance, and was changed to mycophenolic acid (MMF) 2 grams daily; In addition, prokinetics are indicated for digestive involvement.

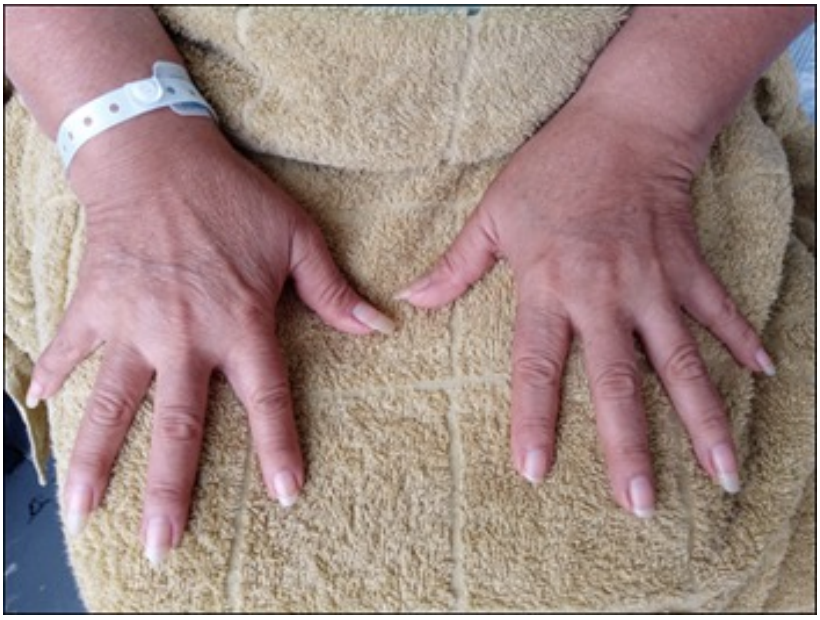

Figure 3. In addition to Raynaud's phenomenon, there was no evidence of proximal or distal skin involvement in both hands.

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The patient begins progressive improvement of the pulmonary and digestive symptoms. Currently, it is with present progressive improvement. Maintains MMF 1 gram daily and weight gain.

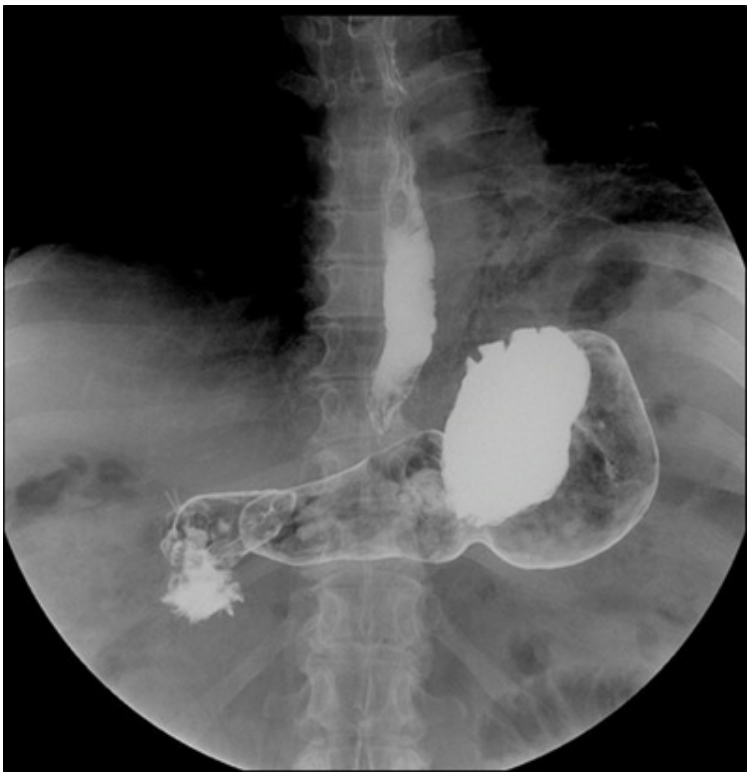

Figure 4. A contrasted study that verifies alteration with slowing of the emptying of the contrast material in the gastric fundus; the patient reported early fullness.

DISCUSSION

Unlike the bibliographic references reviewed (where SSss is classified as a variant of SSlc and positivity necessary for AAC), our patient had SS sine scleroderma, but as a variant of the diffuse cutaneous form, since she presented positivity for AAT and disease. associated pulmonary interstitial [4].

Three types of SSss have been proposed: type 1 (total absence of skin involvement), type 2 (absence of skin sclerosis, but presence of telangiectasias, calcifications, or scars) and type 3 (late-onset skin involvement after visceral involvement)[5]. The patient started the disease with respiratory symptoms; therefore, it is type 1. Taking into account that there was a delay of approximately two years in the definitive diagnosis of SSss in the patient; said wait agrees with the average time (2.5 + 1.5 years)[6].

The immunosuppressant of choice in PSS is methotrexate (MTX), with azathioprine and mycophenolate being valid options. This changes in the SSss, since there is no skin involvement and lung involvement (as was the case with our patient) the drugs of choice are AZA or MMF. In a patient with SSss and general involvement or vital compromise, cyclophosphamide is preferred. Corticosteroids are almost banned in all varieties of PSS (including its sine form)[7].

The prognosis of patients with SSss is good (similar to the limited cutaneous form), however, this largely depends on the time it takes to diagnose and the organ affected [8]. The patient has pulmonary and digestive interstitial disease, but the good response to MMF favors her and she is considered to have a good prognosis. SSlc also has another rare variety called CREST syndrome, where skin involvement occurs in the form of characteristic calcinosis[9].

CONCLUSIONS

In a patient with elevated ANA, absence of skin involvement, dysmotility of the digestive tract, and interstitial lung disease, antibodies for PSS should be requested and prompt and early diagnosis and treatment of SSss should be obtained.

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Authorship contributions: Freddy Liñán Ponce executed the conception, data collection, writing, data analysis and bibliographic search, Juan Leiva Goicochea and José Chávez Corrales approved and approved the final version.

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Correspondence: *Freddy Liñán Ponce.*
Address: *Calle Antúnez de Mayolo N.° 225, urbanización Los Granados, Trujillo, Perú.*
Telephone number: *991 555 068*
E-mail: ***droctavio77@gmail.com***

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