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Systemic Sclerosis: A Hard Approach in a Hardened Skin

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Abstract

Systemic sclerosis (SSc) is a multisystem disease characterized by widespread vascular dysfunction and progressive fibrosis of the skin and internal organs. SSc's prevalence ranges from 30 to 240 per million inhabitants, being females mostly affected. This case is about a female, 43 years old. Visited family doctor with complaints of fatigue, lumbago at nighttime and systemic myalgia. Pain, edema and thumb's paresthesia persisting for a week preceding the time of the appointment. Physical examination showed edema of the distal phalanges bilaterally. No skin or nail changes were present. No signs of arthritis. Raynaud's phenomenon with coarse speckle >1:160, normal ds-DNA and RF, sedimentation rate of 32 mm/h. The patient was referred to a rheumatology consultation. During one year of follow-up in rheumatology consultation, just kept arthralgia without other symptoms. In September 2014, the physical exam showed skin wrinkling in the distal ends of the fingers with positive Raynaud. New immunological study ANA, rheumatoid factor (RF), C3, C4, anti-centromere antibody (ACA), topoisomerase I (anti-Scl-70) and nailfold capillaroscopy were requested. Positive ANA, negative ACA and anti-Scl-70. Nailfold capillaroscopy showed dilated capillary loops, microhemorrhages and architectural derangement. Systemic sclerosis (SE) was diagnosed. Methotrexate and prednisolone treatment were initiated. Currently, progressive worsening of tissue fribrosis despite maximal immunosupression with methotrexate. Tocilizumad treatment under consideration. Annual follow-up with pulmunary function testing (PFT), lung CT and Doppler echocardiography.

Overlapping symptomatology with other diseases such as systemic lupus erythematosus (SLE), dermatomyositis and rheumatoid arthritis may occur. Therapy requires a systemic and multidisciplinary overview of the patient. It must be coherently adapted to target its manifestations, as to improve quality of life and prevent, when possible, disease progression.

Keywords: Systemic sclerosis; Scleroderma; Family medicine; Raynaud phenomenon

Introduction

Systemic sclerosis (SSc) is a multisystem disease characterized by widespread vascular dysfunction and progressive fibrosis of the skin and internal organs [1]. SSc is generally subdivided into limited (lcSSc) and diffuse (dcSSc) cutaneous subsets. Patients with lcSSc typically have skin involvement distal to the elbows and knees, and may display features of the CREST syndrome (calcinosis cutis, Raynaud phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia). Patients with dsSSc generally have skin involvement extending to the proximal limbs and/or trunk and are at a greater risk for the development of significant renal, lung, and cardiac disease [1-7].

SSc should be suspected in patients with skin thickening, puffy or swollen fingers, hand stiffness, and painful distal finger ulcers. Symptoms of Raynaud phenomenon and gastroesophageal reflux are often present [1,3].

The 2013 classification criteria for SSc were developed by a joint committee of the American college of rheumatology (ACR) and the European league against Rheumatism (EULAR). Incorporate disease manifestations of the three hallmarks of SSc: fibrosis of the skin and/or internal organs, production of specific autoantibodies, and evidence of vasculopathy [8,9].

The differential diagnosis in SSc includes scleroderma, scleromyxoderma, overlap syndromes, endocrine disorders, nephrogenic systemic fibrosis, amyloidosis, eosinophilic fasciitis, chronic graft-versus-host disease (GVHD), drug-induced scleroderma and environmental exposures [1,3,10,11].

All patients require symptomatic treatment and both limited and diffuse cases should be treated for vascular manifestations. Early dcSSc requires immunosuppressive treatment. In all cases of SSc vigilante follow up to determine significant organ based complications is mandatory [3,7,10].

Methods

Interview/family evaluation with patient

Results

Personal background

- Female, 43 years old, married, public transport driver, stage 1 of Duvall cycle, middle class according to Graffar scale, Apgar score corresponding to highly functional family.
- Pathological background: esophagitis, hiatus hernia and dyslipidemia.
- Surgical background: oophorectomy and tonsillectomy.
- No medicine or food allergies known.
- Up-to-date vaccination.
- Denied smoking, alcohol or drug abuse.
- No risky sexual behavior

Family background

- Father: psoriasis, diabetes mellitus type II and dyslipidemia.

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- Mother: diabetes mellitus type II.
- Maternal grandmother: Died with colorectal cancer
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Chronic medication treatments

Sinvastatin 20 mg once a day, omeprazole 40 mg once a day, methotrexate 25 mg once a week, prednisone 5 mg once a day, primrose oil twice a day, L-ascorbic acid

Current disease status

- **May 2013:** Visited family doctor with complaints of fatigue, lumbago at nighttime and systemic myalgia. Pain, edema and thumb's paresthesia persisting for a week preceding the time of the appointment. Physical examination showed edema of the distal phalanges bilaterally. No skin or nail changes were present. No signs of arthritis. Raynaud's phenomenon not conclusive. No constitutional symptom. Spine CT, hemogram, sedimentation rate, C-reactive protein (CRP), antinuclear antibody (ANA), anti-double-stranded (ds-DNA), rheumatoid factor (RF), creatine kinase (CK) and serum creatinine level examinations were requested. In case of emergency, treatment with etoricoxib was prescribed.

- **May 2013:** Returns to family doctor with the results of the exams. Overlapping symptomatology. CT shows C3-C5 and C7-D1 degenerative disc disease. Osteophytes in L5-S1 with apparent nerve compression. Analitically, hemoglobin (10.2 g/dL), ANA with coarse speckle >1:160, normal ds-DNA and RF, sedimentation rate of 32 mm/h. The patient was referred to a rheumatology consultation.

- **August 2013:** Evaluated on rheumatology consultation. Patient kept symptomatology. Arthralgia of inflammatory rate. Immunologic study, protein electrophoresis, iron kinetics, serology, chest X-ray and electromyography were requested.

- **October 2013:** No finger swelling, diffuse arthralgia of inflammatory rate. Exams show ANA (1/160 coarse speckle), anti-cyclic citrullinated peptide (anti-CCP) and negative RF. Electromyography shows bilateral mono-neuropathy by compression of the median nerve at the carpal tunnel. No changes observed in the other exams. Orthopedic consultation requested. Etoricoxib treatment showed no improvement. Therapeutic trial with Diprophos® prescribed.

- **March 2014:** Patient claimed transient improvement with Diprophos® but maintained complaints of hands paresthesia and excessive sweating at nighttime. Tuberculosis screening was requested.

- **September 2014:** Persisting symptomatology. Negative tuberculin exam. The physical exam showed skin wrinkling in the distal ends of the fingers with positive Raynaud. New immunological study ANA, RF, C3, C4, anti-centromere antibody (ACA), topoisomerase I (anti-Scl-70) and nailfold capillaroscopy were requested. Anti-RNA polymerase III antibody was not requested due to exam unavailability.

- **November 2014:** Positive ANA, negative ACA and anti-Scl-70. Nailfold capillaroscopy showed dilated capillary loops, microhemorrhages and architectural derangement. Systemic sclerosis (SE) was diagnosed. Methrotrexate and prednisolone treatment were initiated. Pulmonary function testing (PFT), lung CT, Doppler echocardiography, upper gastrointestinal endoscopy (UGI), esophageal manometry and 24 h pH monitoring was requested.

- **February 2015:** Worsening symptoms upon return to consultation. Skin thickening of the hands, face and forearms, perioral skin tightening with decreased oral apertur, digital pitting, and tendon friction rubs of the fingers and ankles and dyspahia with gastroesophageal reflux. Normal PFT and lung CT, pulmonary artery systolic pressure (PASP) 32 mmHg. Gastroesophageal reflux disease confirmed.

- **Currently:** Progressive worsening of tissue fibrosis despite maximal immunosuppression with methotrexate. Tocilizumad treatment under consideration. Annual follow-up with PFT, lung CT and Doppler echocardiography.

Conclusion

Systemic sclerosis is characterized by a heterogeneous phenotype, affecting various organs, rendering differential diagnosis challenging. The diagnosis is based on the presence of physical symptoms and changes of specific antibodies. Overlapping symptomatology with other diseases such as systemic lupus erythematous (SLE), dermatomyositis and rheumatoid arthritis may occur. Therapy requires a systemic and multidisciplinary overview of the patient. It must be coherently adapted to target its manifestations, as to improve quality of life and prevent, when possible, disease progression.

In this particular case, diagnosis was challenging, with several hypothesis having been considered. The verification of the Raynaud's phenomenon led to the complementary set of exams which allowed the final diagnosis of diffuse systemic sclerosis.

References

1. Christopher P (2015). Denton Overview and classification os scleroderma disorders.
2. Dumoitier N, Lofek S, Mouthon L (2014) Pathophysiology of systemic sclerosis: State of the art in 2014. 43(10): e267-e278.
3. Fett N (2013) Scleroderma: Nomenclature, etiology, pathogenesis, prognosis, and treatments: Facts and controversies. Clin Dermatol 31: 432-437.
4. Mouthon L (2014) Systemic sclerosis: Views and thoughts for the future. 43(10): e265-e266.
5. Varga J (2016). Overview of the clinical manifestations of systemic sclerosis (scleroderma) in adults.
6. Christopher P Denton Overview and classification os scleroderma disorders.
7. Denton CP. Overview of the treatment and prognosis of systemic sclerosis (scleroderma) in adults.
8. Johnson SR (2015) New ACR EULAR guidelines for systemic sclerosis classification. Curr Rheumatol Rep 17: 32.
9. van den Hoogen F, Khanna D, Fransen J, Johnson SR, Baron M, et al. (2013) classification criteria for systemic sclerosis: an American College of Rheumatology/European League against Rheumatism collaborative initiative. Arthritis Rheum 65: 2737-2747.
10. Kowal-Bielecka O, Landewé R, Avouac J, Chwiesko S, Miniati I, et al. (2009) EULAR recommendations for the treatment of systemic sclerosis: a report from the EULAR Scleroderma Trials and Research group (EUSTAR). Ann Rheum Dis 68: 620-628.
11. Khanna D (2011) Diagnosis and treatment of systemic and localized scleroderma. Expert Rev. Dermatol. 6: 287-302.