Juvenile dermatomyositis with scleroderma features - when skin thickening is not systemic sclerosis

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Systemic sclerosis (SSc) is an autoimmune disease which main clinical feature is skin thickening. However, there are other rare conditions that may act as mimickers of this skin mark and differential diagnosis is essential for disease management and treatment. Dermatomyositis (DM), an autoimmune muscle disease with a heterogenous spectrum, may be one of those cases, rarely presenting features of SSc which may puzzle the diagnosis[1,2].

A thirty-year-old male, evacuated from Guinea Bissau, was admitted to the Rheumatology inward due to progressive skin thickening and mechanic polyarthralgias with years of evolution. When asked, he also reported muscle weakness since childhood. Physical examination revealed significant skin thickening distal to the metacarphalangeal (MCP) joints (modified Rodnan skin score 6/51)[3] and sclerodactyly of all hands' fingers, both suggestive of SSc (Figures 1 and 2). However, signs of DM were also observed, with skin depigmentation over the MCP and interphalangeal joints similar to Gottron papules, and over the elbows and knees suggestive of Gottron sign, together with generalized muscle atrophy (but with normal muscle strength) (Figures 3 and 4). Blood analyses, namely inflammatory markers and muscle enzymes, were unremarkable, except for a positive anti-NXP2 antibody, a major myositis-specific autoantibody. Paraneoplastic syndrome was excluded after exams and no signs of other organ involvement, such as lung, or Raynaud phenomena were observed. A muscle and skin biopsy were suggested to the patient, but he refused the procedures. Considering all of this, juvenile DM was assumed with features of SSc (not meeting clinical or classification criteria for SSc diagnosis)[4]. Methotrexate 15mg/week and prednisolone 5mg/day were started as an attempt to ameliorate muscle and joint pain, as well as skin thickening, but without improvement of the lesions or pain. Consequently, they were both suspended and no other immunosuppressors were considered as it was assumed that the complaints were a result of disease damage. The patient started physical therapy with a major improvement of the muscle atrophy and joint pain. However, he eventually abandoned the hospital and therefore therapy.

This case reinforces the difficulty of diagnosing rare conditions, especially with overlap features and a long-time progression of symptoms. This patient showed only signs of damage after an active disease during childhood, which made it more of a challenge to diagnose. Some authors suggest giving the name "scleromyositis" to patients who fulfil the criteria for both diagnosis, which is an interesting entity although still without any appropriated treatment[5]. In this case, the patient did not have a clinical diagnosis of SSc, and that is why the authors assumed the diagnosis of juvenile DM with SSc features. Moreover, the delay in diagnosis made the immunosuppressant treatment ineffective, since only damage of the disease was present with no present activity.

Figures

Figure 1 - Dorsal view of both hands of the patient - note the sclerodactyly and the depigmentation over the metacarpophalangeal and interphalangeal joints

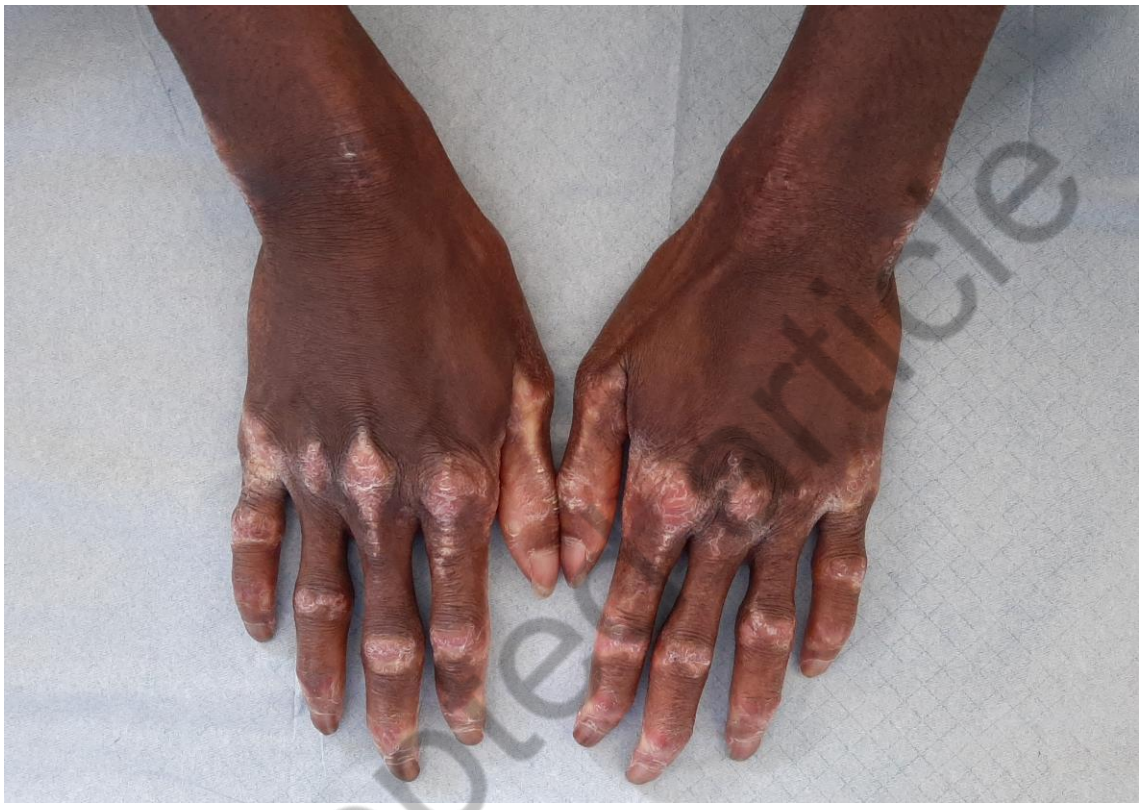


Figure 2 – Palmar view of the left hand of the patient - note the sclerodactyly of all fingers

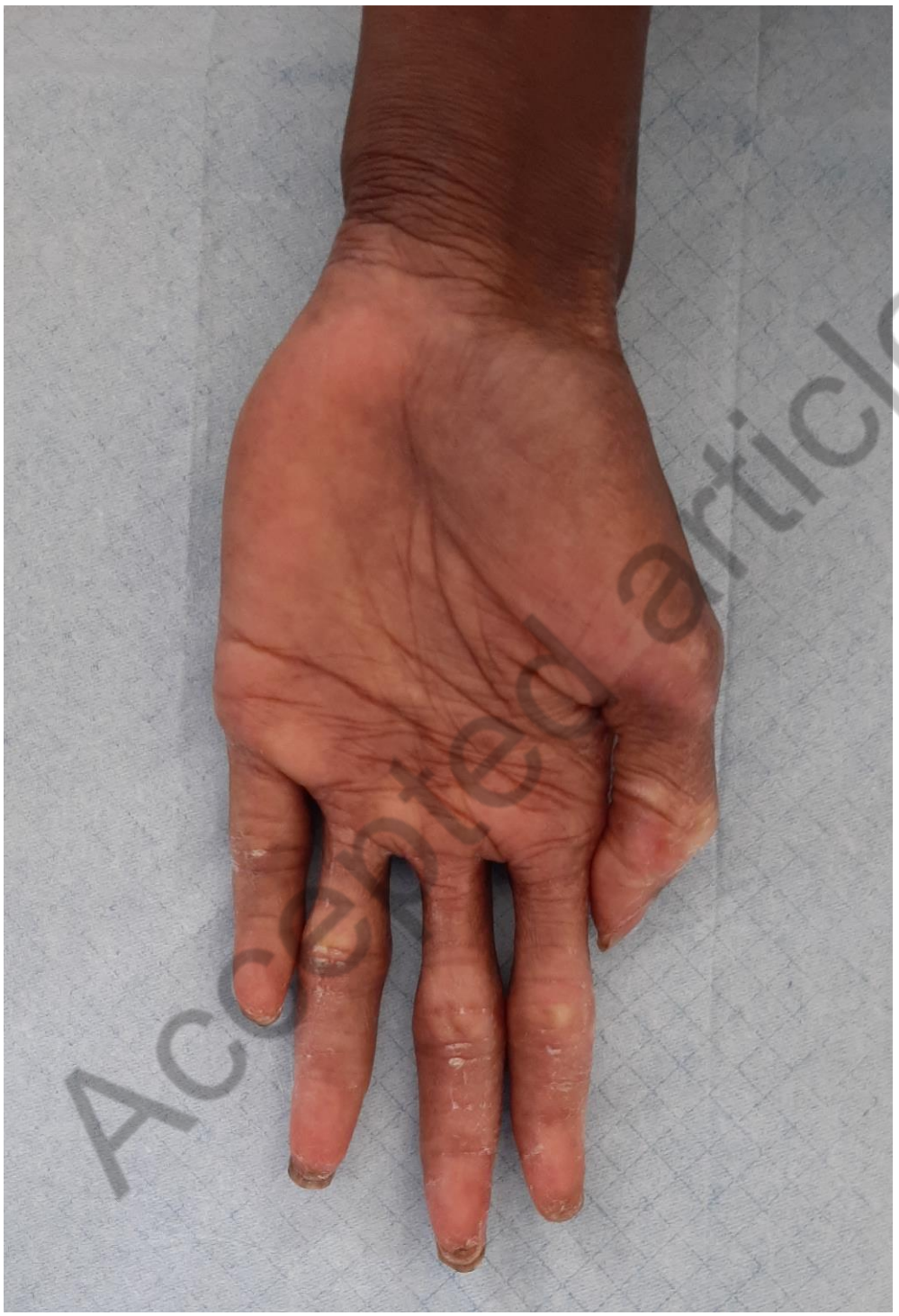


Figure 3 – Posterior view of the right elbow of the patient - note the skin depigmentation over the elbow

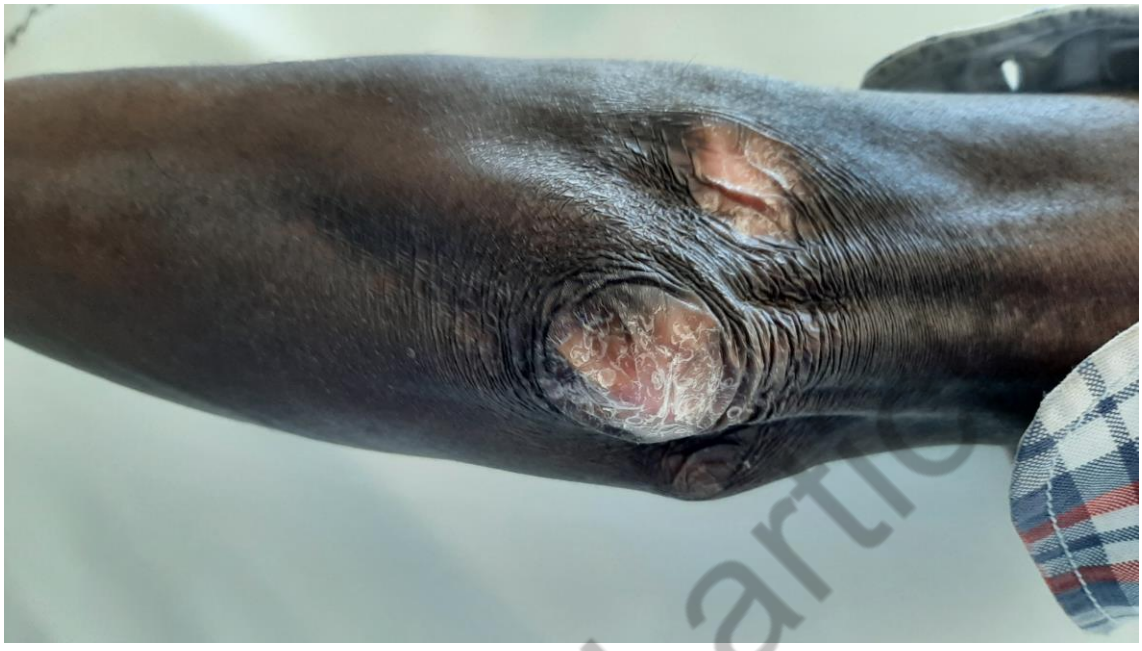
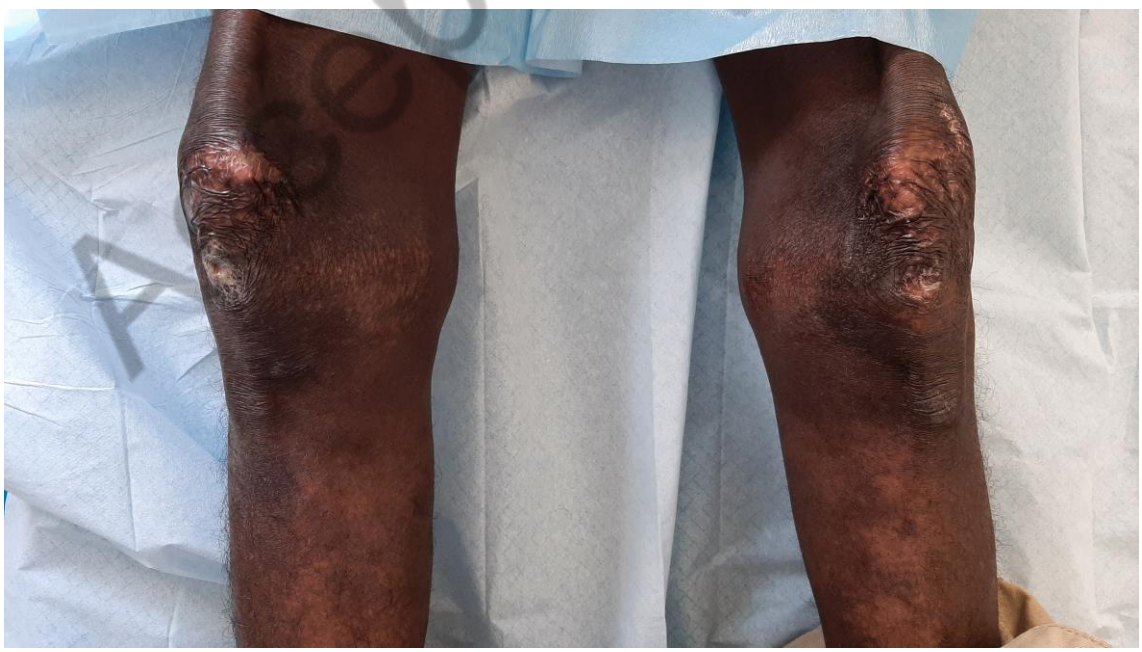


Figure 4 – Anterior view of the knees of the patient - note the skin depigmentation over these joints and the muscle atrophy



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