Case report

A cosmetologist with systemic sclerosis

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Abstract

Scleroderma is a rare disease. This case report highlights its occurrence in a cosmetologist raising the possibility of exposure to organic solvents as a cause as well as the myriad of clinical presentations in such patients. The diagnosis was made using the 2013 ACR/EULAR classification criteria for scleroderma. The essential features were those of widespread hypo and hyperpigmented ('salt and pepper') skin lesions, healed digital ulcers, proximal myopathy, gastrointestinal manifestations, extensive skin fibrosis and tendon friction rub. She tested positive to anti nuclear antibodies with a nucleolar pattern and antibodies to Scl-70 was positive. Her lung function test revealed a restrictive pattern.

Keywords: Scleroderma, Cosmetologist, 'salt and pepper appearance'

Introduction

The first convincing description of scleroderma was in 1753 by Carlo Curzio[1]. He described a 17 year old patient as having 'extensive tension and hardness of skin all over her body'. It was in the mid 19th century that scleroderma was established as a clinical disease entity and given its current name[2].

It is a rare autoimmune connective tissue disease of unknown aetiology characterized by thickening of the skin caused by accumulation of collagen and by injuries to the smallest arteries. A myriad of factors such as genetic, environmental, vascular, autoimmunologic and microchimeric factors are involved in its pathogenesis. According to the 2013 ACR/EULAR classification criteria for scleroderma[3], skin thickening of the fingers extending proximal to the metacarpophalangeal joints is sufficient for a patient to be classified as having scleroderma. If this is not present, seven other additive items are considered, with varying weights for each. This includes finger tip lesions, telangiectasia, abnormal nail fold capillaries, pulmonary hypertension and/or interstitial lung diseases, Raynaud's phenomenon and serological markers such as anticentromere, anti-topoisomerase 1 and anti-RNA polymerase 3 antibodies. Patients with a total score of ≥ 9 are classified as having definite scleroderma.

The incidence and prevalence of Systemic Sclerosis (SSc) varies in different populations. It seems to be more prevalent in United States (276 cases per million adults[4], than in Europe (8-15 cases per million adults)[5]. The annual incidence of new cases has been reported as 1 to 20 cases per million[4,5]. It is three times more common in women than in men[6]. The higher incidence of scleroderma among blacks has been attributed to the postulation that in this ethnic group certain connective tissue responses which are involved in protection against infection and repair after injury may also predispose to certain diseases[6].

Environmental factors could be classified as occupational (silica, organic solvents), infectious (bacterial, viral), and non-occupational/non-infectious (drugs, pesticides, silicones)[7]. Exposure to silica through various occupations remains one of the main environmental risk factors for SSc. Other occupational agents, such as epoxy resins, welding fumes and hand-arm vibration, have been investigated, but no definitive associations may be made due to small sample sizes[8].

Solvents are liquids that dissolve a solid, liquid or gas. Organic Solvents (OSs) are compounds whose molecules contain carbon. Common uses for OSs are: dry cleaning (e.g., tetrachloroethylene), paint thinner (e.g., toluene, turpentine), nail polish removers and glue solvents (acetone, methyl acetate, ethyl acetate), spot removers (e.g., hexane, petrol ether), detergents (citrus turpenes), perfumes (ethanol), nail polish and chemical synthesis[9]. Reinl[10] was the first to point out the association between systemic sclerosis and exposure to solvents.

Exposure to certain organic compounds such as vinyl chloride monomers, trichlorethylene, benzene, and other solvents has been reported as a risk factor of SSc in case reports and in two case–control studies[11, 12]. A meta-analysis published by Aryal *et al*[13] confirmed a significant positive association between exposure to solvents and systemic sclerosis.

Marie *et al*[14] in a study on environmental risk factors for SSc noted a marked correlation has thus been found between SSc onset and occupational exposure to crystalline silica and the following organic solvents: white spirit, aromatic solvents, chlorinated solvents, trichloroethylene, and ketones.

Based on the rarity of this disease, we report a case highlighting its multisystemic presentation and its occurrence in a cosmetologist.

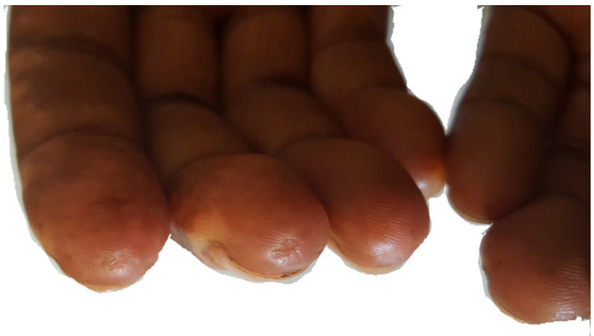
Case report

The patient was a 34 year old single lady who was in stable health until 18 months prior to initial presentation when she noticed skin discoloration involving her right hand. She subsequently developed swelling of both feet and face. The facial swelling was worse in the mornings. There was an associated history of difficulty with breathing on activity but not at rest. There was subsequent affectation of the skin of the face, ears, neck, chest, upper and lower limbs as well as multiple joint pains. She experienced early morning stiffness of an hour's duration. She had digital ulcers which healed leaving atrophic scars. This also involved the dorsal aspects of the proximal interphalangeal joints of the middle digits. She did not experience Raynaud's phenomenon. She complained of occasional fatigue, pruritus, difficulty with standing from a sitting position as well as difficulty with lifting her upper limbs. She had no history of hair loss, oral ulcers or difficulty with swallowing but experienced epigastric pain which radiated to the back. She has had recurrent diarrhoea alternating with constipation with occasional fecal incontinence. She was a cosmetologist who had been in business for a year before onset of symptoms. She was involved in hair fixing, nail polishing, fixing or removal which led to exposure to organic solvents such as benzene sulphate, cholesterol tea-tree oil, olive oil hair mayonnaise, phenoxyethanol, polyanionic cellulose, isopropyl alcohol and propylene glycol. No history of use of botulinum injection for her clients.

Essential examination findings were widespread hypo and hyperpigmented ('salt and pepper') skin lesions on the face, trunk and limbs (Figure 1). She had bilateral pitting pedal edema up to the distal third of the legs and microstomia.

Figure 1: Salt and Pepper appearance in a young lady with systemic sclerosis'

Figure 2: Healed digital ulcers in a young lady with systemic sclerosis



She was tachypneic and the only significant finding in the abdominal examination was fibrotic skin over the lower abdomen. There was loss of skin pluckability extending to the elbows, with extensive fibrosis on the upper and lower limbs worse in both forearms, atrophic scars on the dorsal aspects of the middle digits of the proximal interphalangeal joints, contractures at the elbow joints and tendon friction rub at both ankle joints. She was neither hypertensive nor diabetic.

Her laboratory results revealed a positive anti nuclear antibody with a nucleolar pattern. Antibodies to double stranded DNA, human immunodeficiency virus and rheumatoid factor were negative. Her antibodies to topoisomerase 1 (Scl-70) was positive (>320U/ml). Spirometric findings were in keeping with restrictive lung disease (FEV1 43% predicted). Liver function tests, serum proteins, urinalysis, electrolyte, urea and creatinine were normal. The full blood count revealed anaemia (11.7g/dl), microcytosis (59.3fl) and leucocytosis ($19.8x10^9$). Her chest X-ray revealed cardiomegaly while a pelvic scan showed right adnexal cyst .

Discussion

The reported case met the ACR/EULAR 2013 criteria[3] for definite systemic sclerosis. Systemic sclerosis is subdivided into limited cutaneous scleroderma and diffuse

cutaneous scleroderma. Limited cutaneous scleroderma affects the skin, the fingers, hands, face, lower arms, and legs and may present with CREST syndrome (Calcinosis, Raynaud's phenomenon, Esophageal dysmotility, Sclerodactyly, Telangiectasia). In diffuse cutaneous scleroderma, skin thickening begins in the hands with subsequent involvement of the face, upper arms, upper legs, chest with internal organ affectation such as the lungs, kidneys, stomach and intestines. The higher frequency of the diffuse form in blacks is confirmed by the index case as well as increased occurrence of anti topoisomerase[15]. The female preponderance seen in other reports is highlighted in this case as well as the younger age at presentation amongst blacks. The patient was aged 34 years as against 36.1 years in the series by Tikly *et al*[16] and 51.5 years among Caucasians[17]. She had features suggestive of proximal myopathy. Muscle weakness is a significant problem in SSc and often has more than one cause[18]. A myopathy can occur from a direct extension of the fibrosis into the muscle itself in which case the patient presents with weakness, fatigue with mildly elevated creatine phosphokinase and abnormal electromyography[19].

The patient had a history of digital ulcers with healed atrophic scars (Figure 2). She however did not report having Raynaud's phenomenon which is rarely reported in Black Africans[20]. There was a co morbidity of peptic ulcer disease as well as constipation alternating with diarrhoea and fecal incontinence. Gastrointestinal tract involvement is almost universal in patients with systemic sclerosis and is characterized by abnormal motility secondary to dysfunctions caused by abnormal innervations, smooth muscle atrophy and tissue fibrosis[21]. In a study conducted to determine incidence of gastrointestinal manifestations in patients with systemic sclerosis, Wielosz *et al*[22] reported that gastrointestinal (GI) symptoms were observed in 74% of patients and that upper GI symptoms were observed in 54 (74%) patients and lower GI symptoms in 22 (30%) patients.

She had florid cutaneous manifestations: the 'salt and pepper' appearance, hide bound skin as well as flexion contractures of the digits as well as the elbows. Skin thickening typically peaks in the first 3 to 5 years[23]. This is within the time frame the patient presented and also accounts for the contractures at the digits and elbows.

The patient had microcytic anaemia. Elkayam *et al*[24] reported two patients with systemic sclerosis who presented with microcytic hypochromic anaemia and were found to have watermelon stomach. This could have accounted for the iron deficiency anaemia and its manifestations such as fatigue. A positive anti nuclear antibody with a nucleolar pattern is in consonance with a similar finding amongst three of the nine tested patients in a study by Adelowo *et al*[20]. The presence of antibodies to topoisomerase correlates with the clinical finding of the diffuse type and the presence of interstial lung disease.

The aetiology of systemic sclerosis is unknown. However evidence suggests the role played by environmental factors[13]. Exposure to organic solvents such as benzene might have predisposed her to this condition. Further studies will be required to determine a causal relationship.

Conclusion

Systemic sclerosis is a rare disease. Exposure to organic solvents may trigger the disease in a genetically susceptible individual. The limitations here include unavailability of facilities to carry out investigations such as nailfold video-capillaroscopy as well as the cost of investigations taking into cognizance the fact that most patients are not covered by health insurance and so bear the cost of management.

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