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A Case of Localized Scleroderma in a Sculptor and His Wife

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SUMMARY

The etiology of localized scleroderma is unknown, and its pathogenetic relationship to its systemic counterpart is unclear. Environmental exposures, notably to silica dust, have long been suspected in the pathogenesis of the disorder. However, its relationship to the localized variant has not been well described. Here we present two cases of localized scleroderma in a sculptor and his wife who have extensive exposure to silica dust.

INDEXING TERMS

Localized scleroderma; Morphea; Silica

CASE REPORT

Husband

A 79-year-old man, full-time ceramic sculptor, presented to our clinic for evaluation of progressive morphea, diagnosed in 2004. The patient's initial manifestations included medallion-sized lesions in the bilateral axilla that were indurated and yellowish in color. Rapidly thereafter, he developed increasing numbers of firm indurated plaques that produced a band-like tightening on his abdomen, which diminished his appetite and impaired his ability to breathe. His disease progressed to involve his upper thighs and lower legs, limiting his ability to walk. The patient was initially evaluated by an outside dermatologist, and treated for two months with clobetasol, calcipotriene, and oral prednisone. In addition, he received twice-weekly psoralen and Ultraviolet-A (PUVA) treatments (64 in total). Despite these interventions, the patient reported that his areas of induration had increased in size and tightness.

The patient's past medical history is significant for prostate cancer for which he received radiation therapy, and hypercholesterolemia. On presentation, he denied symptoms of Raynaud's phenomenon or esophageal reflux. He continues to work full-time as a ceramic sculptor, which has been his occupation for more than 50 years. Notably, pervasive 'fumes' circulate through the living quarters of his home of 20 years due to the location of his kiln in the adjacent garage.

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Laboratory examination revealed negative antinuclear (ANA), anti-scl-70, anti-double stranded DNA (dsDNA), anti-centromere, anti-neutrophil cytoplasmic (ANCA) autoantibodies, negative rheumatoid factor (RF), and negative Lyme titer. On physical examination, the patient was tanned and in no apparent distress. The bilateral upper extremities were diffuse firm, indurated plaques from the dorsal hands to the shoulders (Figure 1). There was extensive tightening and induration of the skin on the abdomen, back upper buttock, and the lower extremities from the hips down to the dorsal feet, with sparing of his face. There was no sclerodactyly or evidence of ischemic changes in his fingers. Nailfold capillaries were normal.

Wife

The 69-year old wife of the previous patient revealed that she too had been diagnosed with morphea in 2001. Married to her husband for 25 years, she reported extensive exposure to the 'fumes' from the kiln. Her initial manifestations included hyperpigmented patches on the dorsum of her arms bilaterally, which did not impair her range of motion. Her skin involvement has slowly expanded but has not required any treatment.

Her past medical history is significant for hypertension and hypercholesterolemia. On presentation, she denied symptoms of Raynaud's phenomenon or esophageal reflux. However, over the past six months, she developed a persistent cough, for which she was evaluated by an outside pulmonologist. To date, her work-up included Positron Emission Tomography (PET), Computed Axial Tomography (CAT), and Pulmonary Function Testing (PFT). Her high-resolution CT scan showed bronchiectasis and interstitial disease predominantly in the middle lobe and lingular segments, although the etiology of the changes is not totally resolved at the current time. Notably, she has a 30-pack-year history of smoking, which she stopped ten years ago.

Routine laboratory examination included normal complete blood cell count, liver, and kidney function. Further testing revealed negative antinuclear, anti-dsDNA, anti-Jo-1, anti-SSA, anti-SSB, anti-Smith, anti-ribonucleoprotein (RNP) autoantibodies, and negative RF. On physical examination, the patient had hyperpigmented patches on the dorsum of her arms bilaterally, with sparing of the rest of her skin (Figure 2). Range of motion at the wrist joint was normal. There was no sclerodactyly or evidence of ischemic changes in her fingers. Nailfold capillaries were normal.

HISTOPATHOLOGY

The husband had a skin biopsy from the left lower chest in 2005, which showed diffusely thickened and eosinophilic collagen bundles within reticular dermis (Figure 3). The eccrine glands were compressed within the sclerotic dermis, and the peri-eccrine adipose tissue was markedly diminished. In addition, a superficial and deep, perivascular, and periadenexal infiltrate of lymphocytes, histiocytes, and plasma cells was present. The wife had a skin biopsy taken from the right forearm in 2001. This biopsy showed similar findings with thickened, intensely eosinophilic collagen bundles throughout the reticular dermis, compression of eccrine glands, loss of follicular structures, and a sparse perivascular lymphoplasmacytic infiltrate. No polarizable foreign material or foreign body granulomas were identified in either biopsy.

DISCUSSION

Localized scleroderma is a poorly understood cutaneous sclerosis that has multiple clinical subtypes.¹ Since its initial description in late 19th century, there have been advances in histopathological descriptions and classifications of this clinical entity, but the etiology

continues to elude investigators. Environmental exposures, immune alterations, microchimerism with autoimmunity, trauma, and *Borrelia* infection have all been suggested as contributing to the pathogenesis of the disease.²⁻⁴ Epidemiological, genetic, familial, and twin studies suggest that scleroderma is not a heritable disease but may arise in genetically susceptible individuals when exposed to environmental triggers.^{1,2} The vast majority of cases are sporadic.

Several environmental hazards have been reported as triggering factors for scleroderma-like disorders.⁵ These include organic solvents, vinyl chloride, bleomycin, polymerizing epoxy resins, and silica.^{5,6} Except for silica, all other substances induce changes that only mimic scleroderma, and are classified as scleroderma-like disorders.⁵ To our knowledge, silica exposure has only been associated with systemic sclerosis and not with localized scleroderma. However, both diseases are thought to represent part of the same pathological process, and therefore a discussion of silica-induced systemic sclerosis is warranted. In addition, it should be emphasized that silica has also been associated with other connective tissue diseases.

Bramwell first drew attention to the possible link between occupational silica dust exposure and systemic sclerosis when he reported “diffuse” scleroderma in nine patients, five of whom were stone masons.⁷ Subsequently, there have been many reports describing the association with either silica exposure or silicosis.⁸ Importantly, silica-associated systemic sclerosis is clinically, immunologically, and serologically indistinguishable from patients with idiopathic systemic sclerosis, which is not true for other substances.⁹

Crystalline silica particles vary in size, but it is those smaller than 1 μm that are the most pathogenic.¹⁰ Silica dust can be inhaled into the lung where it is phagocytosed by alveolar macrophages and transported to extrapulmonary sites.¹¹ Importantly, silica can also penetrate the skin of heavily exposed areas. Silica particles between 1 and 20 μm have been isolated in the skin of the arms of patients with occupational scleroderma by phase contrast and polarizing microscopy.¹² The particles destabilize cell and phagosome plasma membranes, leading to the generation of free radicals and releases of lysosomal enzymes.¹³ Additionally, it is postulated that the phagocytosed particles stimulate the macrophages to release cytokines, notably Interleukin-1 (IL-1) and Interleukin-6 (IL-6), which stimulate fibroblast collagen synthesis.^{12,14} The crystals are toxic to macrophages in a dose dependent manner.¹² Upon death of the macrophages, silica is again liberated, and a perpetual cycle ensues culminating in fibrosis.

We postulate that the silica particles from the clay were dispersed throughout the house, and represent the pathogenetic trigger. Physical examination and serology revealed no evidence of systemic involvement in either patient. The reason why the couple developed limited cutaneous disease is unclear. Potentially, the nature of the clay used and particle size of the silica and/or disbursement mechanism might have been different from those described in other case reports. Such particles may have initiated the disease by depositing in the skin and not entering the lung, accounting for the localized involvement. Accordingly, the sculptors’ diffuse cutaneous involvement is consistent with the more extensive nature of his exposure. Further study is warranted because this observation of silica-associated localized scleroderma might elucidate pathogenetic distinctions between systemic and localized involvement.

Upon presentation, the sculptor was started on methotrexate, given his debilitating involvement. Follow-up three months later demonstrated softening of the plaques, and improved mobility and appetite. The wife’s disease has remained stable, requiring no medical intervention. It is unclear at the current time if the lung problems are related to silica

or a different interstitial lung process. Additionally, we recommended that the couple move the kiln to a different location with proper ventilation. It is unclear whether continued exposure to the silica dust will result in disease progression; however, progression to systemic scleroderma is rare.¹⁵

ABBREVIATIONS & ACRONYMS

PUVA	Psoralen and Ultraviolet-A
ANA	Anti-nuclear antibody
dsDNA	Double stranded DNA
ANCA	Anti-neutrophil cytoplasmic antibody
RF	Rheumatoid Factor
PET	Positron Emission Tomography
CAT	Computed Axial Tomography
PFT	Pulmonary Function Testing
RNP	Ribonucleoprotein
IL	Interleukin

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FIGURE 1.
Sculptor: (A) Indurated skin on chest; (B) Medallion-sized, indurated, yellow plaque on right axilla.

FIGURE 2.

Wife: Faint hyperpigmented patches on the dorsum of arm.

FIGURE 3.

(A) The biopsy shows diffusely thickened and eosinophilic collagen bundles within the reticular dermis, as well as a superficial and deep, perivascular, and periadenexal lymphoplasmacytic infiltrate with scattered histiocytes; (B) The diffusely thickened and eosinophilic collagen bundles are more pronounced upon closer examination.