

Case Report

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TANAFFOS 

When Silica Returns-Erasmus Syndrome, a Rare Entity

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Introduction: Systemic sclerosis (SS) is an autoimmune disease with vascular changes and diffuse tissue fibrosis. Silica exposure is one of the strongest recognized risk factors for the development of systemic sclerosis (relative risk 3.2). The co-occurrence of systemic sclerosis in a patient with silica exposure is termed Erasmus Syndrome. Few case reports are available in the literature.

Case presentation: We report here a case of Erasmus syndrome in a 54-year-old stone worker. The patient presented with diffuse skin thickening, hyperpigmentation, Raynaud's phenomenon, and arthralgia along with radiological features of silicosis. Serological markers of systemic sclerosis were strongly positive, and skin biopsy confirmed scleroderma. Hence, a diagnosis of Erasmus syndrome was made. The clinical, serological, and histopathological features of SA-SS in the patient were indistinguishable from those of idiopathic SS; however, a history of silica dust exposure and evidence of silicosis supported the diagnosis of SA-SS.

Conclusion: Careful screening should be done in patients with silicosis along with systemic manifestations to rule out any associated connective tissue disorder. A detailed exposure history should be carefully obtained for all patients with systemic sclerosis.

Keywords: Systemic sclerosis (SS); Silica-associated systemic sclerosis (SA-SS); Silicosis; Scleroderma

INTRODUCTION

Systemic sclerosis (SS) is a rare immune-mediated connective tissue disease affecting small arteries, microvessels, and connective tissue, characterized by fibrosis and vascular obliteration in the skin and internal organs. Erasmus syndrome (ErS), a rare condition, was first described in 1957 as the association between silica exposure and the subsequent development of systemic sclerosis (SS), with or without concurrent silicosis (1). The prevalence of ErS in SS is reported to be low. We intend to report this rare entity in the following case report.

CASE SUMMARIES

A 54-year-old male, a non-smoker, presented with a history of progressive shortness of breath for the last one

and a half year. He also had progressive skin thickening over the last 4 months; initially, it occurred on the dorsum of the feet and hands, which later started to involve the forearm and trunk, along with scaling over the abdomen and a burning sensation throughout the whole body. He had difficulty in swallowing, especially solid foods, along with regurgitation of food after 10-15 minutes of food intake. He had occasional bluish discoloration of fingers on exposure to cold and arthralgia. Occupational history revealed that he worked as a stone cutter for nearly 27 years in a stone quarry and left the job four years ago. There was no history of hematuria, hemoptysis, or oral ulcers. Neither any family history of similar complaints nor any history of chronic drug administration in the past was noted.

On examination, the patient exhibited sclerodactyly, diffuse skin thickening (Figure 1), hyperpigmentation, and a modified Rodnan skin score of 36, along with prominent telangiectasia, Raynaud's phenomenon, digital pit scars, loss of forehead wrinkles, and small hyperpigmented scales over the abdomen. Examination of the respiratory system revealed bilateral end-inspiratory fine crepitation. Other systemic examination was within normal limits.



Figure 1. Photograph of the patient's volar aspect of hands and fingers showing puffy and thickened skin of fingers

Complete blood counts, urinalysis, liver and renal function tests, and serum electrolytes were within normal limits. HIV, HCV, and HbsAg serology were negative. Antinuclear antibody was highly positive (1:640) with a mixed pattern, and anti-Scl-70 was strongly positive. Chest X-ray showed bilateral diffuse reticulonodular opacities with a conglomeration of nodular opacities in the peri-hilar region (Figure 2). CT chest showed progressive massive fibrotic changes in bilateral upper lobe posterior segments, multiple tiny foci of calcifications with few non-calcified nodules and reticular opacities bilaterally; and mediastinal lymphadenopathy with internal punctate foci of calcifications (Figure 3). The patient had a severe restrictive defect with an FEV1 of 1.52 L (45% of the predicted) and a FEV1/FVC ratio of 83.5. USG of the whole abdomen and pelvis revealed a normal study, and a single contrast esophagogram was also normal. Echocardiogram showed left ventricular ejection fraction of 60%, mild TR (RVSP 36mmHg), and trivial MR. Using clinical judgment, serological and radiological findings, a diagnosis of

Erasmus syndrome was made. Later skin biopsy from the right side of the abdomen showed thickened dermis, composed of broad sclerotic collagen bundles with minimal chronic inflammatory infiltrate suggestive of scleroderma, which confirmed our diagnosis.

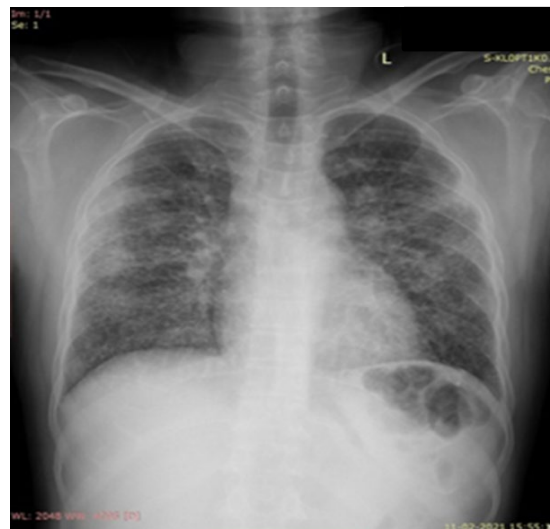


Figure 2. Chest skiagram showing bilateral diffuse nodular opacities with confluent opacities over the bilateral upper zone

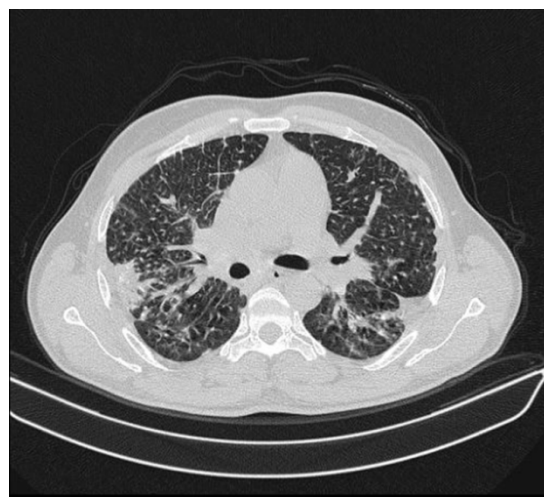


Figure 3. CT thorax shows bilateral upper lobe perilymphatic nodules and fibrotic areas

DISCUSSION

Silicosis is probably the most common form of pneumoconiosis, characterized by irreversible lung fibrosis that develops from prolonged pulmonary inhalation and retention of crystalline silica, along with an immune reaction mounted by the body in response to this foreign

chemical. It mainly occurs as an occupational hazard in individuals engaged in stone quarrying, mining, and sandblasting (2). The frequency of occupational exposure to silica dust is also recognized as a risk factor for many other systemic autoimmune diseases, including rheumatoid arthritis, systemic lupus erythematosus, and small-vessel vasculitis with renal involvement (3).

Understanding the relation between environmental risk factors and the development of SS is challenging. This may be due to the phenotypic and pathogenic heterogeneity of patients and disease, and also to the poor capability to quantify environmental exposure and to assess the role of gene-environment interactions in this disease (4). The risk of developing SA-SS after exposure to silica dust is up to four times greater than that for the general population (5). The dysregulation of T lymphocytes by exposure to silica is the most credible theory explaining this association (6,7). The silica particles are phagocytized by macrophages and transported to regional lymph nodes. The stimulated macrophage produces cytokines (IL1, 2, 6, TNF- α , TGF- β , and other cytokines), which constitute a permanent stimulus for fibroblast activation, which increases their production of collagen and glycosaminoglycan, leading to cutaneous sclerosis, vascular occlusion, and pulmonary fibrosis (5,8,9). The same (IL1, IL2, IL6, and TNF) and possibly other cytokines also induce the maturation and activation of dendritic cells in the regional lymph nodes. It follows a polyclonal activation and proliferation of native T cells. Such T cells are thought to operate to cause vascular occlusion and cutaneous sclerosis (10). The generation of anti-topo I antibodies (the predominant autoantibodies present in silica-associated SS) in genetically susceptible individuals may depend partly on the patient's sex and on the site of organ involvement and may be triggered by silica particles acting as an immune adjuvant (11).

The prevalence of ErS in SS is 0.3 to 0.9% of overall SS and 43% of male SS (12,13). It has a worldwide distribution and affects all races; however, epidemiological data from Asia, particularly India, are lacking (14). The first case of Erasmus syndrome in India was reported by Khanna et al.

in a 53-year-old stone-cutter (15). Since then, a few case reports have been published about the disease (14,16-24), and three of these cases are reported from Rajasthan (14,19,24). Among these, two have reported the coexistence of pulmonary tuberculosis and Erasmus syndrome (16,23).

The clinical, serological, and histopathological parameters of SA-SS in our patient were indistinguishable from those of idiopathic SS, but the patient had a history of exposure to silica dust and features of silicosis, which led us to the diagnosis of SA-SS. Hence, careful screening should be done in patients with silicosis along with systemic manifestations to rule out any associated connective tissue disorder. Most of the cases described in the literature occurred in patients with occupations that involve an intense exposure to silica dust (12,13), which is consistent with our finding. However, less intense exposures may also play a role in ErS development (7, 25). So, an exposure history should be sought carefully in all patients with systemic sclerosis.

Several occupational hazards, particularly exposure to silica dust, have been documented in the literature as triggers for the development of systemic sclerosis. Therefore, primary prevention should involve the use of personal protective equipment for at-risk professionals.

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