International Journal of Science and Healthcare Research
Vol. 8; Issue: 4; Oct.-Dec. 2023
Website: ijshr.com
Case Report *ISSN: 2455-7587*

Erasmus Syndrome: A Rare Case Report of Systemic Sclerosis with Silicosis and Progressive Massive Fibrosis

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DOI: https://doi.org/10.52403/ijshr.20230410

ABSTRACT

Erasmus Syndrome is a rare disorder that is characterized by the development of systemic sclerosis after significantly prolonged exposure to silica with or without silicosis. Systemic Sclerosis is a multisystem autoimmune disease with vascular changes, inflammation, and diffuse tissue fibrosis. We report a case of Erasmus syndrome in a 45-year-old male who worked in the marble industry. The patient presented with difficulty in breathing, skin tightening, joint pain, Raynaud's phenomenon, microstomia, and progressive massive fibrosis.

Keywords: Erasmus Syndrome, Systemic Sclerosis, Silicosis, Progressive Massive Fibrosis

INTRODUCTION

The concept of Erasmus Syndrome was given by Erasmus in 1957 who first described scleroderma in 17 gold miners from Witwatersrand.[1] Erasmus syndrome is a rare occupational rheumatological disorder in which systemic sclerosis develops following exposure to silica, with or without silicosis. Silicosis term was coined by Visconti in 1870.[2] Silicosis, the most common pneumoconiosis is a preventable occupational inflammatory disease of lung characterized by irreversible lung fibrosis which develops after prolonged inhalation and retention of crystalline silica and immunological reaction. Systemic sclerosis is a complex immunological, clinical heterogenous progressive disease having inflammation and degeneration of skin, heart, lung, kidney, and gastrointestinal tract.[3] It is characterized by organ-specific autoantibodies, end-organ fibrosis, and small vessel vasculopathy.

CASE PRESENTATION

A 45-year-old non-diabetic, normotensive male presented with complaints of increased shortness of breath, cough with expectorant, and low-grade fever from 1 year. Then developed skin tightening, bilateral symmetrical polyarthritis, bluish discoloration of fingertips & toes on exposure to cold, digital ulcerations, hair loss & dyspepsia from 6 months. The occupational history revealed that he worked in the marble industry for nearly 5 years & then was forced to leave his job 15 years back due to breathlessness. Two months ago the patient was discharged with the diagnostic query of silico-tuberculosis from some tertiary hospital where bronchoalveolar lavage was taken which was sterile & GeneXpert for tuberculosis was negative. There is also a past history of anti-tubercular treatment 10 years ago. No history of any chemotherapeutic drug. No relevant family history. On examination, the vitals were stable with oxygen saturation of 94%, pallor, microstomia, grade 2 clubbing,

skin thickening with modified Rodnan Score of 24, sausage-shaped fingers (Fig. 1), areas of irregular hyperpigmentation on arms, back, and legs s/o salt and pepper dermopathy (Fig. 2) was present. The respiratory examination revealed bilateral basal crepitation in inter and infra scapular areas and decreased breath sounds in the right lung. The chest X-ray revealed bilateral large reticulo-nodular opacities with calcified hilar nodes. (Fig. 3) High-resolution computed tomography thorax showed irregular soft tissue mass with consolidation s/o progressive massive fibrosis (PMF) in the right upper and apical-posterior segment of the left upper lung lobe with multiple calcified nodular infiltrates, interlobular septal thickening, eggshell calcified mediastinal lymph nodes & dilated lower oesophagus. The ANA was strongly positive (74.24 AU/ml) with a positive Anti-Scl-70 antibody (2.68 OD ratio). Pulmonary function test revealed a severe restrictive pattern with forced vital capacity of 30% of predicted. Sputum was negative for acid-fast bacilli and cartridge-based nucleic acid amplification testing (CBNAAT). Echocardiography, complete blood count, liver and renal function tests were normal except for mild normocytic normochromic anaemia. Treatment was made with low-dose oral prednisolone, cyclophosphamide, nifedipine, pentoxifylline, inhalational bronchodilators, and doing good on follow-up.

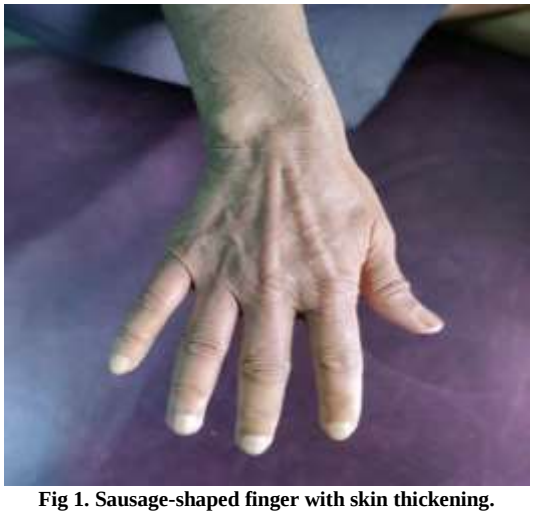

Fig 1. Sausage-shaped finger with skin thickening.

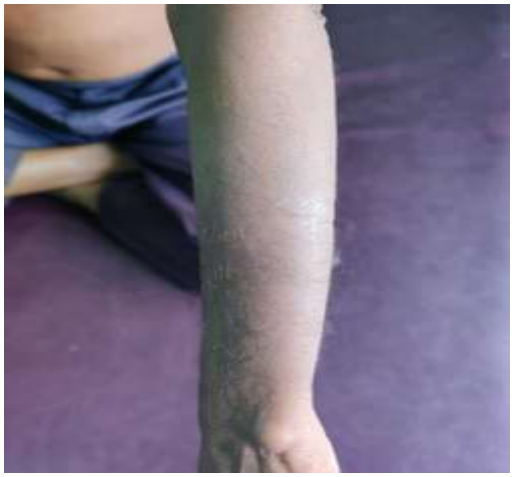

Fig 2. Salt and paper dermopathy

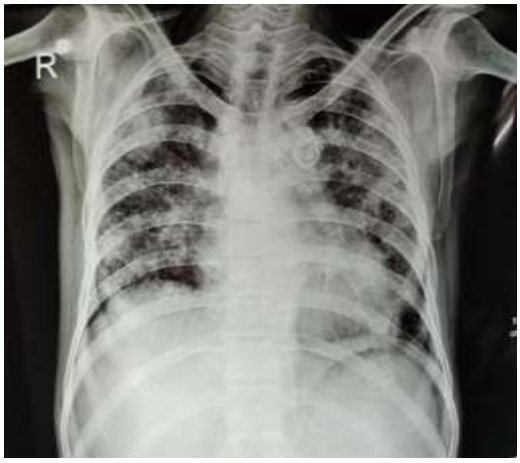

Fig 3. Plain radiograph chest, posteroanterior view showing bilateral large reticulo-nodular opacities (right > left) with calcified hilar nodes.

CASE DISCUSSION

Systemic sclerosis is a chronic inflammatory complex clinical heterogenous orphan disease with protean clinical manifestation involving variable degrees of collagen accumulation in affected tissues and obliterative vasculopathy. It has a frequently progressive course and significant disability, disfigurement, and mortality. Raynaud's phenomenon, telangiectasias, pulmonary artery hypertension, interstitial lung disease, skin thickening, and esophageal dysmotility are the cardinal manifestations. The associated autoantibodies are anti-centromere, anti-topoisomerase I (anti-SCL70), and anti-RNA polymerase III antibodies. Systemic sclerosis is female predominant disease with a 3 to 7:1 female-to-male ratio while the Erasmus syndrome is

male predominant which may be due to increased direct occupational exposure.[2,3] Silicosis occurs in persons working at processing silica-containing stones, like stone cutters, mining, granite quarrying, and sandblasting. Many environmental and occupational exposures are implicated in the pathogenesis of autoimmune diseases like rheumatoid arthritis, systemic sclerosis. A lot of environmental factors are extensively analyzed for associations with systemic sclerosis like silica, vinyl chloride, toluene, benzene, epoxy resins, rapeseed oil, drugs, pesticides, breast implants (silicones) etc.[4] Silica exposure is a major risk factor for other diseases like pulmonary tuberculosis, lung malignancy, autoimmune diseases like rheumatoid arthritis, systemic lupus erythematosus, systemic sclerosis. The exact pathophysiologic mechanism is not very clear. The continuous inhalation of silica leads to the deposition of silica in terminal bronchioles and alveoli, initiating an inflammatory cascade leading to macrophage activation, upregulation of cell-signal pathways, increased cytokines, free radicals' generation and increased levels of inflammatory mediators like Interleukin-1, Interleukin-2, Tumor necrosis factor. These factors lead to fibroblast proliferation and collagen deposition.[2] The continuous silica exposure leads to abnormal humoral and cellular immunity, hypergamma-globulinemia, altered T-helper and T-suppressor lymphocytes and is thought to be a cause of progressive systemic sclerosis.[6] Silica exposure also mediates epigenetic changes like DNA methylation and histone modification.[4] The silicosis can be acute, chronic, or accelerated. International classification of Radiograph of Pneumoconiosis considers any pulmonary opacity more than 1 cm as progressive massive fibrosis on chest X-ray.[5] The small nodules confluent together to form large masses or opacities over time. Along with this, the calcification of perihilar and mediastinal lymph nodes known as eggshell calcification can be present. These evolve into more complex diseases and large fibrous masses. Erasmus syndrome is much less reported in India. Ganguly. J., et al 2013 reported a similar case who had diffuse parenchymal lung disease due to silicosis along with all clinical and immunological features of systemic sclerosis.[7] Goyal. A., et al 2013 reported a case of Erasmus syndrome with acid-fast bacilli positive pulmonary tuberculosis.[8] Sharma R.K. et al 2020 reported a similar case of systemic sclerosis with silicosis with positive bronchoalveolar lavage gene expert for acid-fast bacilli.[9] Silicosis-induced or silica-associated systemic sclerosis cannot be distinguished from idiopathic systemic sclerosis either clinically or immunologically although they have higher and severe lung involvement along with anti-Scl-70 antibody.[10] The risk is higher in miners as compared to non-miners with silica exposure.[11] Lomante, J.M.J., et al. 2022 reported a case of a Filipino man who developed Erasmus Syndrome after working in a grind mill for 10 years with a brief literature review of 20 patients reported in the last 10 years.[2]

However, the reported incidence is very low considering the number of workers involved in silica-based occupations. So, there must be other genetic and environmental factors playing a role that need to be identified. Secondly, lots of patients remain undiagnosed and the exact occupational relevance is not well known. So, proper knowledge and awareness should be created to prevent this disorder

The presence of clinical manifestations like skin thickening, arthralgia, microstomia, Raynaud's phenomenon, and serological markers of systemic sclerosis, progressive massive fibrosis with significant exposure to silica, diagnosis of systemic sclerosis with silicosis i.e., Erasmus syndrome was made.

CONCLUSION

Erasmus syndrome is a rare occupational rheumatological disorder in which systemic sclerosis develops following exposure to silica with or without silicosis.

Crystalline silica exposure from stone quarrying, mining, or marble workers not only causes silicosis but also autoimmune diseases like systemic lupus erythematosus, rheumatoid arthritis, systemic sclerosis, and vasculitis.

Systemic sclerosis is a multisystem disease characterized by organ-specific autoantibodies, end-organ fibrosis, and small vessel vasculopathy.

Idiopathic or systemic sclerosis associated with silicosis are indistinguishable clinically or immunologically.

Declaration by Authors

Acknowledgment: None

Source of Funding: None

Conflict of Interest: The authors declare no conflict of interest.

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How to cite this article: Hargurdas Singh, Nishant Kumar Singh. Erasmus syndrome: a rare case report of systemic sclerosis with silicosis and progressive massive fibrosis. *International Journal of Science & Healthcare Research.* 2023; 8(4): 67-70. DOI: *https://doi.org/10.52403/ijshr.20230410*

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