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RAYNAUD'S PHENOMENON AND PERICARDIAL EFFUSION REQUIRING MANAGEMENT WITH A PERICARDIAL WINDOW AS THE ONLY MANIFESTATIONS OF SYSTEMIC SCLEROSIS WITHOUT SCLERODERMA FOR OVER 10 YEARS, AFTER WHICH PULMONARY HYPERTENSION DEVELOPED

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ABSTRACT

Introduction: Systemic sclerosis (SSc) is a rare autoimmune connective tissue disease characterised by vasculopathy and progressive fibrosis affecting the skin and internal organs. Systemic sclerosis sine scleroderma (ssSSc) is an uncommon variant, accounting for 5–10% of cases and is defined by the absence of cutaneous involvement despite internal organ manifestations.

Case description: We present a case of a patient who initially presented with recurrent pericardial effusion and late-onset Raynaud's phenomenon. Despite serological markers, SSc was not diagnosed, due to absence of skin involvement, until pulmonary hypertension (PH) developed over a decade later. Nailfold capillaroscopy confirmed ssSSc. Surgical pericardial drainage and targeted PH therapy led to clinical improvement.

Discussion: Pericardial effusion may be an early and under-recognised manifestation of ssSSc. While typically attributed to inflammation, our patient had a transudative effusion with a poor response to steroids, suggesting a non-inflammatory mechanism. The effusion preceded PH by over a decade, challenging current pathophysiological theories. Surgical drainage was effective, contrasting with poorer outcomes in cutaneous SSc.

Conclusion: This case highlights the diagnostic complexity and heterogeneity of cardiac involvement in ssSSc. It underscores the importance of maintaining a high index of suspicion in patients with unexplained cardiopulmonary symptoms and demonstrates the indolent course of the disease in atypical SSc.

KEYWORDS

Systemic sclerosis, systemic sclerosis sine scleroderma, pericardial effusion, pulmonary hypertension

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LEARNING POINTS

- Systemic sclerosis sine scleroderma (ssSSc) may be diagnosed late due to the absence of skin involvement, underscoring the need for clinical vigilance.
- Visceral manifestations can precede Raynaud's phenomenon, challenging the traditional diagnostic sequence in systemic sclerosis.
- Pericardial effusion may be an early and under-recognised feature of ssSSc, often presenting with non-specific symptoms and requiring a high index of suspicion.

INTRODUCTION

Systemic sclerosis (SSc) is a rare, multisystem autoimmune disorder characterized by vasculopathy and fibrosis in the skin and internal organs[1]. Systemic sclerosis sine scleroderma (ssSSc) is an uncommon variant of the condition. It can be diagnosed using the Poormoghim criteria, which includes the absence of skin thickening, the presence of Raynaud's phenomenon (RP) or other peripheral vascular involvement, positive antinuclear antibodies (ANA), involvement of one visceral organ typical of SSc and the absence of another defined connective tissue disease[2]. There is conflicting evidence in the literature regarding the prevalence of this condition, with estimates ranging from 5–10% of SSc cases[1,3]. This variability is likely attributable to diagnostic uncertainty, particularly in patients without skin involvement[4]. Patients are either misdiagnosed with idiopathic fibrosis of the affected organ, or they are later reclassified[3]. One study demonstrated that more than 50% of ssSSc patients were later reclassified as having limited cutaneous SSc after a mean follow-up of 1.9 years[3]. Despite this diagnostic variability, a consistent finding across the literature is the marked female predominance, with over 80% of patients being women[3,4].

The most common antibodies associated with ssSSc are the anti-centromere and the anti-topoisomerase I (scl70) antibodies[2,4]. RP is reported as the most common initial presentation of the condition, often preceding visceral symptoms by several years or occurring concurrently[3,4]. Rarely, as in the case of our patient, it may present after the onset of other disease manifestations[4]. While a few case reports describe pericardial effusion preceding cutaneous involvement in SSc, the literature regarding its occurrence as the initial presentation of confirmed ssSSc remains sparse[5,6]. We report a case where pericardial effusion was the initial manifestation of ssSSc, though its clinical significance was only recognised retrospectively following the development of PH.

CASE DESCRIPTION

A 68-year-old Afro-Caribbean woman presented with a 10-year history of late-onset RP, which began in 2014. Her past medical history was significant for recurrent large-

Investigation	Results
Pericardial fluid analysis	Total WBCs 0.2, RBCs N/A, polymorphonuclear neutrophils N/A, mononuclear cells N/A, gram stain and culture N/A, LDH 293 U/l (reference range <450 U/l) Histology: The smears include a mixture of cells which consists of lymphocytes, histiocytes and a great number of very reactive mesothelial cells, both singly and in monolayer groups different in shape and size. A cell block and immunohistochemical studies were performed. Cytological features and immune profile favour the diagnosis of a reactive process.
ANA	Positive speckled Sp100 pattern, positive centromere pattern, ANA titre 1:640 throughout.
ENA	Anti-centromere positive, Anti-scl70 positive
cANCA, pANCA	Negative
Anti-CCP antibodies	Negative
Anti dsDNA	Negative
Antiphospholipid antibody screen	Negative
Serum protein electrophoresis	No monoclonal paraprotein

Abbreviations: WBC, white blood cell; RBC, red blood cell; N/A, not applicable; LDH, lactate dehydrogenase; ANA, antinuclear antibody; ENA, extractable nuclear antigen; ANCA, anti-neutrophil cytoplasmic antibody; Anti-CCP, anti-cyclic citrullinated peptide; DNA, deoxyribonucleic acid; Anti-ds DNA, anti- double stranded DNA.

Table 1. Results of pericardial fluid analysis and autoimmune screen.

volume pericardial effusions starting in 2011, with her only symptom being a persistent productive cough. Notably, she never developed features of cardiac tamponade. Her treatment course included repeated pericardiocentesis, alongside colchicine and a gradually tapered regimen of corticosteroids. Pericardial fluid analysis demonstrated a reactive effusion *(Table 1)*. In the search for an underlying aetiology, serological testing revealed a strongly positive ANA titre (>1:640), along with positive anti-centromere and anti-Scl-70 antibodies *(Table 1)*. These findings led to a diagnosis of idiopathic autoimmune pericarditis. Despite medical therapy, the effusion continued to reaccumulate and was ultimately managed with a pericardial window in 2013. Histological analysis of the tissue sample obtained during the procedure revealed a normal parietal pericardium. Over the subsequent 13 years, there was no significant re-accumulation of the effusion. Throughout this period, she remained under annual rheumatology follow-up due to her serological profile and RP. Clinically, she remained well, and her RP was effectively managed with conservative measures. There were no cutaneous manifestations of SSc, such as sclerodactyly, telangiectasia, or calcinosis. Additionally, there were no sicca symptoms, joint involvement, or any pulmonary or gastrointestinal features suggestive of visceral fibrosis. Consequently, a diagnosis of SSc was not established.

In August 2024, the patient was hospitalised with type 1 respiratory failure and tested positive for Coronavirus disease 2019. Examination was unremarkable. Chest X-ray showed cardiomegaly, electrocardiogram revealed normal sinus rhythm, and bedside echocardiography demonstrated a trivial pericardial effusion, unchanged from the previous year.

Computed tomography pulmonary angiogram excluded pulmonary embolism, but a review of the images by the Respiratory team raised concerns for PH *(Fig. 1)*, which was later confirmed via transoesophageal echocardiogram *(Table 2)*. One-month post-discharge, outpatient cardiac magnetic resonance imaging (MRI) revealed diffuse myocardial fibrosis and right heart strain consistent with PH *(Table 2, Fig. 2)*.

Within two months, she was readmitted with a productive cough, worsening breathlessness, and reduced exercise tolerance. She tested positive for respiratory syncytial virus. On examination, oxygen saturation was 88–90% on room air, with slightly raised jugular venous pressure, bilateral pedal oedema, and a loud second heart sound. No telangiectasia, digital ulcerations, nail fold infarcts, synovitis, or skin thickening were noted. She had no gastrointestinal symptoms of SSc, and high-resolution computed tomography of the chest showed no lung fibrosis *(Table 2)*.

Rheumatology was consulted due to a suspected underlying diagnosis of connective tissue disease. The absence of skin thickening, presence of RP, positive SSc antibodies, autoimmune pericarditis and PH supported a diagnosis of ssSSc. After discharge, the patient underwent nailfold

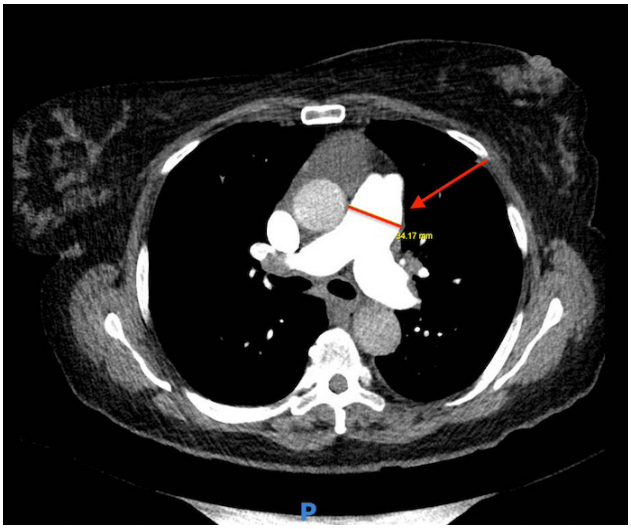


Figure 1. Computed tomography image demonstrates a dilated main pulmonary artery, consistent with pulmonary hypertension. The main pulmonary artery diameter is 34.17 mm as shown by the red arrow.

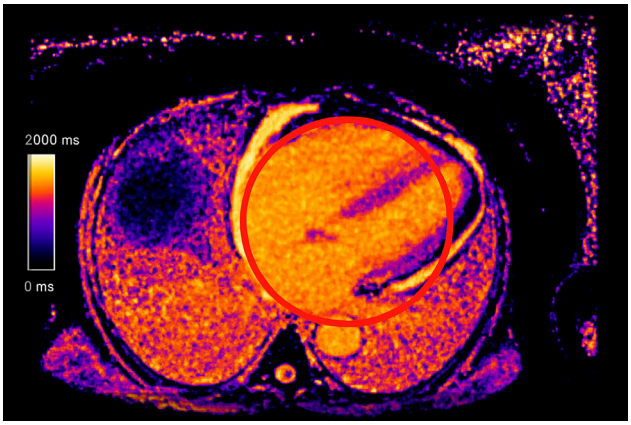


Figure 2. Cardiac magnetic resonance imaging native T1 mapping demonstrating diffusely elevated myocardial T1 values (red circle), suggestive of diffuse myocardial fibrosis.

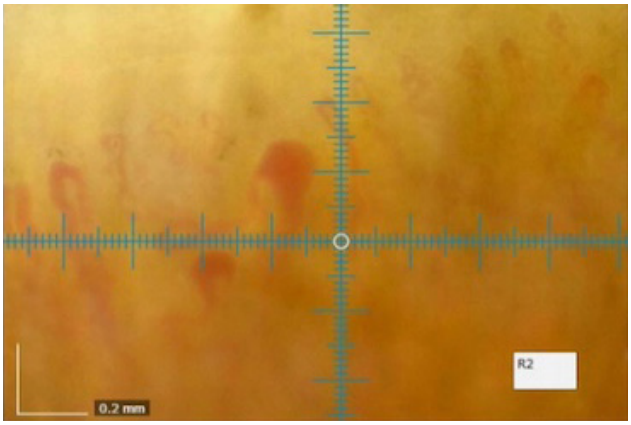


Figure 3. Photographic image of abnormal nailfold capillaroscopy with dilated capillary loops, a giant loop and haemorrhages.

capillaroscopy *(Fig. 3, Table 2)*, which confirmed the diagnosis. She was also referred to the Pulmonary Hypertension Unit for further investigations *(Table 2)* and treatment. She was commenced on sildenafil and ambrisentan, but due to poor tolerance, ambrisentan was replaced with macitentan. Her breathlessness improved significantly with medication and

Imaging	Results
Transoesophageal echocardiogram	There is septal flattening, mild RVH, preserved RV function, TR of velocity of 3.7 ms, small global pericardial effusion. No convincing LVH but there is paraseptal LV band, slight aneurysm or interatrial septum and the features were consistent with precapillary PH.
High resolution computed tomography scan of the chest	Mild centriacinar emphysema. Mild bronchial wall thickening and subtle geographical areas of air trapping bilaterally (on expiratory phase) suggestive of mild small airway disease. No evidence of lung fibrosis. Mild atelectatic parenchymal changes in bilateral lung bases and lingula. No suspicious lung nodule or mass. No focal areas of consolidation to suggest active lung infection. Trachea and main bronchi are patent. No significant axillary or mediastinal lymphadenopathy. Elevated right hemidiaphragm. No pleural effusion. Cardiomegaly noted. Mild pericardial effusion as seen in the previous scan. Marked triple-vessel coronary artery calcification. Normal calibre oesophagus. No aggressive bony lesion.
Cardiac magnetic resonance imaging	Normal LV cavity size and normal EF- 60%. Normal wall thickness and normal LV mass index. Normal left atrium. Mildly dilated RV with mildly reduced RV EF- 46%. Moderately dilated RA. No regional wall motion abnormalities. Flows not reliable hence Qp:Qs cannot be calculated. The difference in stroke volumes between RV and LV is small (11 ml) and likely related to TR. Global pericardial effusion, deepest around RA (12 mm). Pericardium is normal thickness with no clear evidence of pericardial oedema. No previous infarction. Non-specific inferior RV insertion point fibrosis (although this can be seen more commonly in RV pressure overload). No obvious myocardial oedema. Some evidence of more diffuse fibrosis on T1 mapping but this is of uncertain significance.
Incremental shuttle walk	Patient managed 130metres. Oxygen saturations 96% at rest dropping to 83% with exercise, heart rate rising from 72-125 bpm. Blood pressure rose from 143/101 to 171/105 mmHg. She does have Raynaud's in her fingers which may have affected the saturations recording on exercise.
Pulmonary function test	FEV_1 1.13 (66%), FVC 1.57 (76%), FEV_1/FVC 72%. TLCO 2.3 (38%), KCO 54%.
Right heart catheterization	RA 8 mmHg with PA 55/21 mmHg. Mean PA pressure 36 mmHg, PAWP 8 mmHg. Cardiac output 4.67 with cardiac index 2.54 and PVR 480 dynes/sec/cm^2 (*> 2 Wood units), MVO_2 77.2%
Nailfold capillaroscopy	Nail fold capillaroscopy was abnormal with some dilated capillaries on the third finger and a giant haemorrhage seen on the fourth finger of the right hand, again dilated capillaries on the second finger of the left hand.
Serum protein electrophoresis	No monoclonal paraprotein

Abbreviations: RVH, right ventricular hypertrophy; RV, right ventricle; TR, tricuspid regurgitation; ms, milliseconds; LVH, left ventricular hypertrophy; LV, left ventricle; PH, pulmonary hypertension; EF, ejection fraction; RA, right atrium; Qp:Qs, pulmonary to systemic flow ratio; ml, millilitres; FEV1, forced expiratory volume in one second; FVC, forced vital capacity; TLCO, transfer factor for carbon monoxide; KCO, carbon monoxide transfer coefficient; PA, pulmonary artery; PAWP, pulmonary artery wedge pressure; PVR, Pulmonary vascular resistance; MVO_2: mixed venous oxygen saturation.
*2 Wood units = 160 dynes/second/cm^2

Table 2. Results of imaging and other diagnostic investigations.

physiotherapy, and she remains well and continues to be followed up on an outpatient basis.

DISCUSSION

This case illustrates that pericardial effusion can be an early and potentially overlooked presentation of ssSSc. Other cardiac manifestations may include congestive heart failure and arrhythmias[3,4]. Myocardial fibrosis is hypothesised to be the primary driver of most cardiac manifestations in ssSSc[3,4], and notably, our patient's cardiac MRI revealed features consistent with this pathology. While cardiac involvement is a recognised manifestation of SSc, it is less common, affecting approximately 15–25% of patients, compared to gastrointestinal complications (80%) and pulmonary involvement (66%)[4,7]. Only 5-16% of SSc patients exhibit symptoms of pericardial effusion, most of which are non-specific, as with our patient[7,8]. Notably, post-mortem studies have found that pericardial involvement can occur in up to 78% of cases, indicating it often remains undetected during life due to non-specific or absent symptoms[7,8].

The pathophysiology of severe pericardial involvement in SSc remains poorly understood, with considerable variability and inconsistency across the literature.

In one review, Fernández Morales et al.[8] proposed that inflammation is a key factor in pericardial involvement in SSc. Evidence supporting this includes fluid analysis showing an exudative pericardial effusion in 88.9% of cases, indicating an inflammatory process[8]. Histological examinations of pericardial biopsies have also revealed varying degrees of inflammation, such as fibrosis, inflammatory infiltrates, and leukocytoclastic vasculitis, although some biopsies appeared normal[8]. Interestingly, our patient's pericardial

fluid was transudative according to Light's criteria, with a low cell count, normal pericardial biopsy findings, and poor clinical response to corticosteroids. These findings support the hypothesis proposed by Dunne et al.[9] that the pathogenesis of SSc-related pericarditis may not be primarily driven by inflammation. In the context of PH, Dunne et al.[9] also suggested that pericardial effusion could result from haemodynamic factors, such as elevated right atrial pressure impairing lymphatic drainage. However, in our case, the large effusion preceded the onset of PH by more than a decade. Taken together, these contrasting findings illustrate the heterogeneity of pericardial involvement in SSc and underscore the need for further research to clarify its underlying mechanisms. The management of pericardial effusion in SSc typically involves medical therapy with non-steroidal anti-inflammatory drugs, colchicine and corticosteroids[10]. However, medical management proved ineffective in our patient, requiring escalation to pericardiocentesis and ultimately a pericardial window[10]. This favourable outcome following surgical management contrasts with poorer outcomes in cutaneous SSc cases[9], suggesting ssSSc may represent a distinct phenotype, with a more indolent course.

The patient's PH was managed according to established clinical guidelines with a phosphodiesterase-5 inhibitor and endothelin receptor antagonist, with good effect[11].

In our patient, nailfold capillaroscopy played a pivotal role in confirming the diagnosis. According to the classification criteria for SSc, a diagnosis can be established without skin thickening when features such as SSc-specific autoantibodies, RP, lung involvement and abnormal nailfold capillaries are present[3].

This case highlights the need to consider pericardial effusion as a potential early manifestation of SSc. It reinforces the importance of maintaining a high index of suspicion in atypical presentations and calls for further research into the cardiovascular complications of the condition. Timely, multidisciplinary evaluation remains key to accurate diagnosis and effective management.

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