RADIOLOGIC FINDINGS IN SCLERODERMA

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Systemic Sclerosis, aka Scleroderma

Connective tissue disease of unknown etiology

• fibrosis and thickening of skin (most cases) with involvement of internal organs – heart, lungs, kidneys, GI

• Incidence 18-20 per 10^6 people per year. Estimated 75-100,000 cases in USA

• Peak age of onset 30-50, four times more common in women than in men

• heterogeneous group of disorders
Subtypes of Systemic Sclerosis

- **Diffuse cutaneous**
  - skin involved on trunk, face, proximal and distal extremities
  - anti-Scl70 (anti-topoisomerase I) antibody

- **Limited cutaneous**
  - aka CREST (Calcinosis, Raynaud’s phenomenon, Esophageal dysmotility, Sclerodactyly, and Telangiectasias)
  - skin involvement distal to elbow and knee, face, and neck
  - anti-centromere antibody

- **Others**
  - sine scleroderma, “in overlap”, undifferentiated connective tissue disorder
The Cause of Systemic Sclerosis is Unknown

- Genetic contribution: concordance in identical twins 5.9%, HLA linkage
- Silica, metal dusts, and polyvinyl chloride exposures have been linked to increased risk in some studies.
- No conclusive evidence at this time for infectious agent
Fibroblasts, Endothelial cells, and Lymphocytes: Oh My!

Three important pathological themes:

- **Tissue fibrosis** – increased production of collagen and ECM by fibroblasts
- **Inflammation** – macrophage and T cell, more pronounced at earlier stages
- **Microvascular disease** – intimal proliferation, vessel narrowing and thrombosis

It’s not clear what the initiating event is. See excellent Jimenez review for several models.
Many Organ Systems May Be Affected in Systemic Sclerosis

<table>
<thead>
<tr>
<th>Pulmonary</th>
<th>Leading cause of mortality and major morbidity. Interstitial lung disease, pulmonary hypertension.</th>
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<tr>
<td>GI</td>
<td>Esophageal disorders especially common. Stomach and bowel may be affected.</td>
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<td>Renal</td>
<td>“Scleroderma renal crisis”—sudden onset of malignant hypertension and renal insufficiency</td>
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<td>Cardio</td>
<td>High prevalence of arrhythmias, EKG abnormalities</td>
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<tr>
<td>Musculoskeletal</td>
<td>Arthralgia, myopathy, acro-osteolysis, osteopenia, subcutaneous calcinosis</td>
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And others, too.
Z. is a 32 year old woman who immigrated to the US from China 3 years ago. She was initially referred for workup of Raynaud’s phenomenon.

In addition to her fingers “turning blue,” she has noticed increased dyspnea with exertion over the last year. Z. also reports coarsening of the skin on her fingers but attributed this to her work in a restaurant kitchen.

She denied heartburn, reflux, dysphagia, or other symptoms.

On exam, she was found to have mild edema of her hands, digital ulceration, and skin thickening to her mid-forearms. She did not have any calcinosis cutis or telangiectasias.
Patient Z. - 2

Z. had faint basilar inspiratory crackles, greater on the right. She did not have any signs of pulmonary hypertension.

Her resting oxygen saturation was 100% on room air. She was able to walk up 8 flights of stairs without stopping, although she did desaturate to 90% and became mildly dyspneic.
Patient Z. - 3

She had a positive ANA at 1:320 (diffuse) and her anti-Scl70 titer was positive. Serologies for anti-centromere, anti-Sm, anti-dsDNA were all negative.

Pulmonary function tests indicated a TLC of 53% predicted and a DLCO of 41% predicted.

A high resolution chest CT was done.
Sections through apical lung look relatively normal. However, note the patent esophagus.
Sections closer to the lung bases show evidence of interstitial lung disease (predominantly sub-pleural in location) with septal thickening, bronchiectasis, and bronchiolectasis.
Note areas of ground glass opacification.
The lungs are most severely affected at the bases bilaterally.
Radiographic Manifestations of Pulmonary Disease in Scleroderma

A. Chest film
   - May appear normal or show only subtle changes
   - Reticular-nodular pattern most often. Localized initially to lung bases, may ultimately involve lower 2/3 of lungs.
   - Pleural effusions uncommon
   - May see signs of pulmonary hypertension

B. CT
   - 44% with “normal” CXR have abnormal high res. CT
   - subpleural lines (74%), honeycombing (30%), parenchymal bands (26%), thickened septae (22%), subpleural cysts (17%)
   - mediastinal adenopathy may be present
Some important points, highlighted by Z’s case

• **Raynaud’s is the presenting complaint** in ~70% of patients with diffuse disease and in virtually all patients who develop CREST

• Patients with diffuse disease are more likely to present with hand and finger edema ("edematous scleroderma"), arthritis, evidence of organ involvement, and skin thickening. May be seen with Raynaud’s or within 1-2 years.
Patient Z. - 5

• **Esophageal pathology** is common in systemic sclerosis in both diffuse and localized disease.

• **Pulmonary involvement** is the leading cause of mortality in systemic sclerosis. Current therapeutic options remain limited but new treatments are being developed.
Several additional interesting images...

The next cases are from different patients and are included here to illustrate other interesting radiologic findings in patients with systemic sclerosis.
Esophageal Disease in Scleroderma

As in other regions of the GI tract, smooth muscle atrophy and fibrosis leads to defective motility.

Patients may therefore experience dysphagia or odynophagia. In addition, stasis predisposes to esophageal candidiasis.

Poor contraction of the LES makes reflux a significant problem for many patients. Reflux may lead to esophagitis or stricture formation for example.
This image from a double-contrast upper GI study done on a 49 year old man with CREST shows:

- dilatation of the distal esophagus
- a patent gastroesophageal junction
- numerous filling defects in the esophagus (shown by the arrow)

Further endoscopic evaluation lead to a diagnosis of esophageal candidiasis.
Small Bowel Disease in Scleroderma

As seen with the esophagous, smooth muscle atrophy and fibrosis impair motility.

Symptoms may include bloating, anorexia, nausea, and vomiting. Malabsorption may occur secondary to bacterial overgrowth.

The small bowel may be dilated and have an “accordion-like” appearance (sometimes likened to a stack of coins) due to fibrosis-related shortening of the bowel.
This image was taken from the double contrast UGI series done on the 49 year old patient with CREST mentioned earlier. Note the close approximation of the valvulae conniventes ("stack of coins" sign) in the proximal small bowel indicated by the arrow.
Subcutaneous Calcinosi

Subcutaneous calcinosis is another common finding.

Affects ~40% of patients with limited disease and a smaller fraction of those with diffuse disease.

Calcium deposits may become inflamed and cause discomfort. They may extrude through the skin and become infected.
This patient had multiple medical problems including scleroderma, dermatomyositis, and osteoarthritis. She had noticed swelling of her third and fourth digits. Note the prominent **soft tissue calcifications** indicated by the arrows.
References


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