SCLERODERMA: An Update

What You Need To Know
The onset of systemic sclerosis is typically:

A. In the 1st decade of life
B. Between the 4th and 6th decade
C. Equal in all age groups
D. After the 7th decade
The following laboratories is required to diagnose scleroderma:

A. ANA
B. Anticentromere antibody
C. SCL 70
D. None are required
Reynaud’s Phenomenon is common in the population with no connective tissue disease, ranging in 3–10 percent.

A. True
B. False
A patient with shortness of breath and abnormal pulmonary exam warrants:

A. PFTs
B. Echo
C. CT chest
D. All of the above
DIAGNOSIS

Localized
• Morphea
• Generalized Morphea
• Linear disease
• Coup al Sabre

Systemic sclerosis
• Limited cutaneous systemic sclerosis (CREST)
• Diffuse cutaneous systemic sclerosis (diffuse scleroderma)
Circumscribed morphea

An indurated plaque with central hypopigmentation and peripheral erythema is present in this patient with circumscribed morphea.

En coup de sabre with secondary alopecia

A shiny, linear, hypopigmented and hyperpigmented sclerotic plaque is present on the forehead and scalp in this patient with en coup de sabre. Note the presence of secondary alopecia.

Generalized morphea on the trunk

Isomorphic, hyperpigmented, sclerotic plaques are present on the chest and abdomen.
Systemic sclerosis

- An uncommon disease: in 300 million population about 20 new cases/million/year
- About 100-150,000 in the US
- Onset: Usually 30-50 years old, rarely under 10
- Female 3–5: 1, increased in African-Americans
- Survival is decreased
  - Past cause scleroderma renal crisis
  - Current cause pulmonary fibrosis and pulmonary hypertension
Scleroderma Diagnosis

• Clinical diagnosis by Rheumatologist
• Raynaud’s
• Skin thickening of the hands/face and/or swollen fingers
• GERD
• Other organs
  – Pulmonary fibrosis
  – Pulmonary hypertension
  – GI, diarrhea, malnutrition
  – Kidney or cardiac involvement
Laboratory Diagnosis

- **Laboratories not required**
  - Most common tests ANA and SCL 70
  - ANA is usually positive but may be negative
  - Anticentromere antibody is associated with limited cutaneous systemic sclerosis (CREST) and a better prognosis
  - Can be false positives, particularly slightly positive results
Clinical features of limited and diffuse scleroderma

• Limited cutaneous
  – Raynaud’s usually first symptom, may occur for many years before diagnosis
  – Puffy fingers
  – Skin thickness limited to the hands, feet and face
  – Anticentromere antibody
  – Milder general/joint symptoms

• Diffuse cutaneous
  – Raynaud’s often delayed
  – Acute onset common with severe constitutional symptoms
  – Arthralgias, CTS, tendon friction rubs
  – Early diffuse skin involvement
  – SCL 70 antibody
CREST acronym

- Calcinosis
- Raynaud’s
- Esophagus (GERD)
- Sclerodactyly
- Telangiectasia
Raynaud’s Syndrome

• Skin color change with cold, emotion, or sometimes temperature change up or down
• No clear criteria, but generally requires white color changes of the fingers
• There may be blue or purple color changes
• Common in the population with no connective tissue disease, 3–10 percent
• Digital tip pits suggest underlying pathology
Raynaud phenomenon

Panel A shows sharply demarcated pallor in several fingers resulting from the closure of digital arteries. Panel B shows digital cyanosis of the fingertips resulting from vasoconstriction in the thermoregulatory vessel in the skin. 

Courtesy of Fredrick M Wigley, MD.
Digital pitting scars in a patient with limited cutaneous systemic sclerosis.

*Courtesy of Carol M Black, MD, FRCP.*
Skin thickening, sclerodactyly

- Characteristic dermal thickening
- Starts in the fingers and progresses centrally
- Key distinction is skin involvement **above** forearm
  - limited disease with a good prognosis – below forearm
  - diffuse disease with a poor prognosis – **above** forearm
Modified Rodnan skin score for systemic sclerosis

Method used to semi-quantify skin thickness in scleroderma. The modified Rodnan skin score is obtained by clinical palpation of 17 different body areas (fingers, hands, forearms, upper arms, chest, abdomen, thighs, lower legs, and feet) and subjective averaging of the thickness of each specific site: 0 = normal (A); 1 = mild (B); 2 = moderate (C); and 3 = severe (D). The maximum total score is 51.

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Puffy hands and shiny skin in early systemic sclerosis

(A) Diffusely puffy hands are a common initial presentation.
(B) Shiny skin suggests impending skin thickening.

Oral manifestations of systemic sclerosis

(A) Perioral skin tightening with decreased oral aperture, furrowing around the lips, and dry membranes.
(B) Periodontal disease with regression of gum and loosening of teeth.
(C, D) Telangiectasias on lips and tongue.

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Telangiectasia

• Can be seen anywhere, but classically is most visible on the face
• Usually starts with or shortly after Raynaud’s appears
Telangiectasias in scleroderma

Telangiectasias in a patient with limited cutaneous systemic sclerosis.

Courtesy of Carol M Black, MD, FRCP.
Calcinosis

• Starts with a solid nodule under the skin
• May progress to an inflammatory “pustule”
• Resolves when a piece of calcium has spontaneously erupted
• Patients usually describe it as a “grain of rice” coming out of the skin
• Avoid temptation to remove surgically
Calcinosis in scleroderma

Calcinosis on the fingertip of a patient with limited cutaneous systemic sclerosis.

*Courtesy of Carol M Black, MD, FRCP.*
Esophageal/GI involvement

• **GERD** - Often trouble swallowing, heartburn, reflux
• **Stomach** - Bloating, inability to eat full meals/need to eat small frequent meals
• **Watermelon stomach** (GAVE)
• **Intestine** - Diarrhea, malabsorption, bacterial overgrowth, constipation, rectal prolapse, fecal incontinence
Scleroderma, bad stuff

- Scleroderma renal crisis
- Pulmonary hypertension
- Pulmonary interstitial disease
- Malnutrition due to GI disease
Scleroderma renal crisis

- Patients with diffuse skin disease in the first year
- Sudden onset of severe hypertension. Often in the 200/140 range
- Worsening renal function, acute
- Rarely occurs without hypertension
- Captopril is life-saving
- Have your scleroderma patients check their blood pressure daily
Pulmonary hypertension

- Usually in patients with limited scleroderma (CREST) after 8-12 years
- Often a long asymptomatic period with gradually increasing pulmonary pressures -- suddenly become symptomatic and progress to death quickly if not treated
- Routine screening can detect early disease
- Early treatment results in much better outcomes
Pulmonary fibrosis

- Usually in patients with diffuse skin disease
- Basilar crackles
- Subtle changes on chest x-ray only
- CT chest reveals interstitial changes with “groundglass” and fibrosis
- Treatment is difficult, but has been shown to improve disease
• CREST, common diagnostic features of disease
• Limited skin disease = good prognosis
  – No skin thickening above the forearm except face
  – Often positive anticentromere antibody
• Diffuse skin disease = guarded prognosis
• Scleroderma patients check blood pressure daily -- Captopril is life-saving
• Lifetime yearly monitoring of PFTs and echo to detect pulmonary hypertension
• Pulmonary hypertension must be confirmed with right heart cath
• SOB, abnormal pulmonary exam: should obtain PFTs, echo and CT chest
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