

Group of autoimmune diseases with ↑fibroblast activity:

Localised - CREST syndrome now called limited cutaneous systemic sclerosis (lcSSc) .

Systemic affecting internal organs - diffuse cutaneous systemic sclerosis (dcSSc).

Transitory (dcSSc/lcSSc), systemic scleroderma sine scleroderma, & malignant forms rarer.

Epidemiology

- Uncommon. Incidence 2 per 100,000 per year.
- Age of onset usually ~30-40.
- 3F:1M

Risk Factors

- Genetic predisposition.
- Environmental triggers: organic solvents, adhesives, or coatings, viruses.

Presentation

Non-specific: fatigue, musculoskeletal pains and hand swelling.

Raynaud's phenomena followed by hardened skin: common to both lcSSc & dsSSc

Symptoms and signs of lcSSc (formally CREST syndrome)

- Calcinosis of skin (40%)
- Raynaud's phenomena (90-95%)
- oEsophageal involvement (90%) resulting in progressive dysphagia and dyspepsia
- Sclerodactyly (90%) tight, shiny digit skin, if banded → "coup de sabre" appearance.
- Telangiectasia (50%) on face, lips tongue, chest wall etc.

In addition may have:

- Non pitting oedema of the fingers
- "Salt and pepper" appearance from areas of hypo-/hyperpigmentation
- Dilated capillary loops at the base of the fingernails
- Microstomia from tightening of the skin produces "puckered lips"
- Ulceration of skin
- Involvement of internal organs may occur

Symptoms and signs of dcSSc

- Worst in the first 3-5yrs after onset then plateau or some improvement.
- Any of the lcSSc features and may also have: pruritis, arthritis, myositis, weakness, SOB, scleroderma affecting heart, kidneys, lungs, GI tract or other internal organs

Diagnostic Criteria

Features of the disease are divided into 2 groups:

- Major features: centrally located skin sclerosis (arms, face, and/or neck).
- Minor features: sclerodactyly, erosions, atrophy of fingertips, & bilat lung fibrosis.
- Diagnosis of systemic sclerosis = 1 major and 2 minor criteria.

Investigations

Bloods: ESR, FBC (Hb↓ microangiopathic haemolysis, plt↓), CK↑ (myositis), UEC, RF (33% +ve)

Serology: Abs to - centromere (lcSSc), Scl-70 (dcSSc), topoisomerase (dcSSc). ANA (90%)

Skin biopsy: may be useful.

Screening for organ involvement: CXR/chest CT/lung fn tests (lung), hand x-ray (calcinosis, osteopenia), ECG/Echo (heart), endoscopy/barium studies (GI strictures), renal USS.

Management

Supportive:

- Counselling, heated clothing for Raynaud's.
- Avoid cold, smoking and trauma.
- Physiotherapy & occupational therapists

Specific disease altering drugs: None.

Immunomodulation:

- Corticosteroids, **colchicine**, immunosuppressives may be beneficial.

Treat complications:

- Pruritis -antihistamines/emollients
- Raynaud's - CCB
- Myalgia and arthralgia - NSAIDs
- Renal involvement - ACEI
- GORD - PPIs
- Lung fibrosis - **cyclophosphamide**
- Pulmonary HT - endothelin antagonists

Trialled Rx include:

- Autologous stem cell transplantation
- Anti-fibrosis drugs (**bosentan**, **INFγ**, **relaxin**).

Surgery:

- Calcinosis nodules
- Skin ulcers
- Contractures may require surgical intervention.

Complications

- Pulmonary hypertension with fibrosis or bronchiectasis is common and can be fatal.
- Renal crisis (accelerated HT, oliguria, headache, fatigue, oedema, and ↑↑Cr)
- Heart failure from myocardial or pericardial disease, arrhythmias.
- Skin and joint complications include contractures and ulceration.
- GI problems include obstruction, malabsorption, incontinence, rarely perf/peritonitis.
- High risk of miscarriage, early delivery and complications of pregnancy.
- ?Increased risk of malignancies (breast, ovary, oesophagus, colorectal, multiple myeloma, lymphoma)

Prognosis

- 10yr survival is 71% for lcSSc and 21% for dcSSc
- Severe problems normally occur <5yr of diagnosis
- Modern management of pulmonary hypertension is improving the prognosis.