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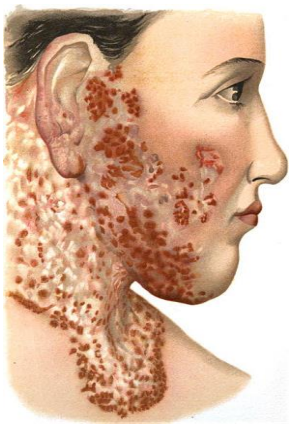
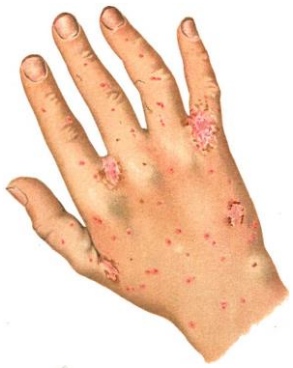
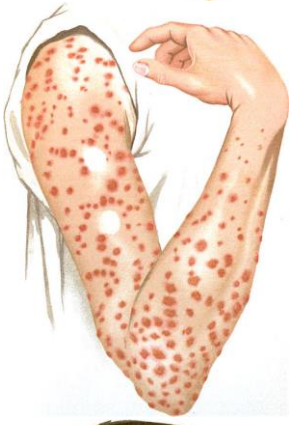
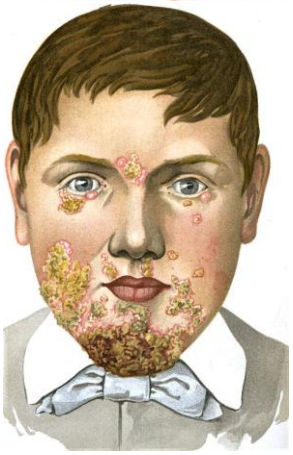


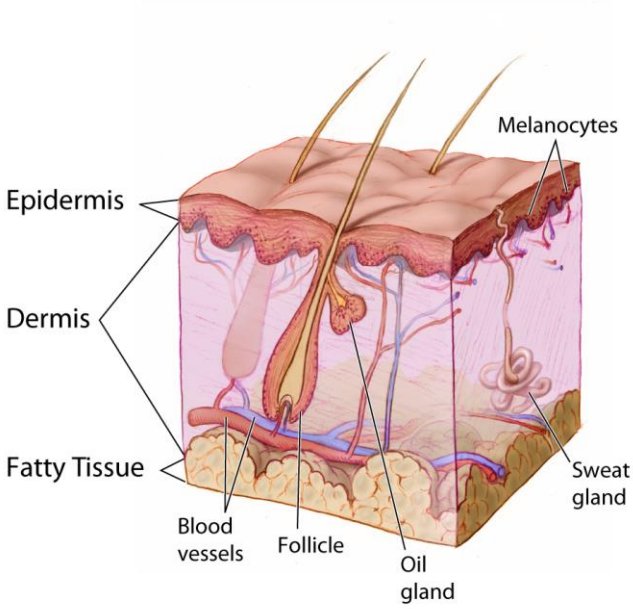






432 Teams

Dermatology



Connective Tissue Diseases

Color Code: Original, Team's note, Important, Doctor's note, Not important, Old teamwork

Done by: ***Fahad AlShayhan***

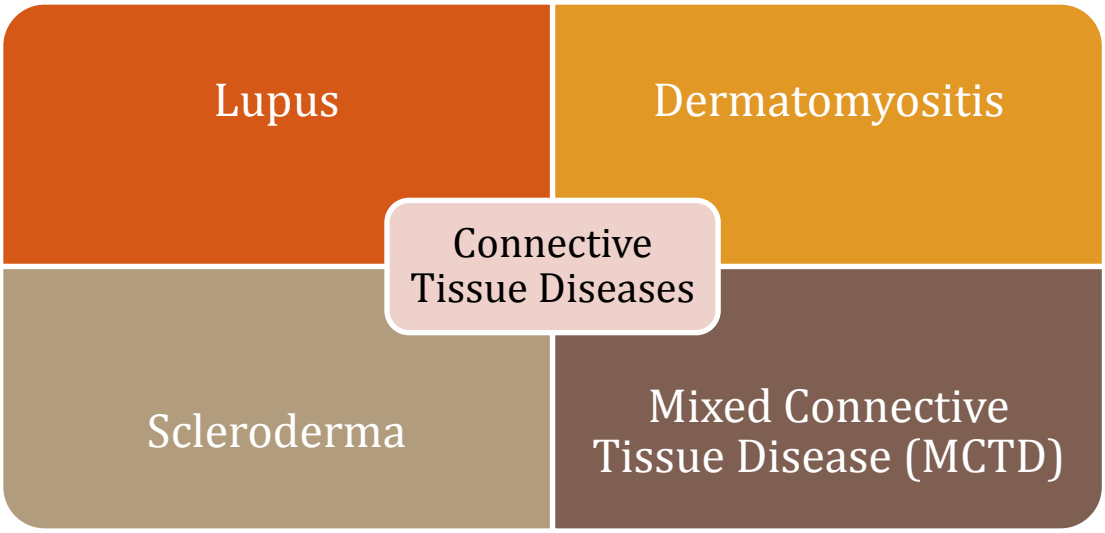
Reviewer ***Yasser al Rumih***

Team Leader: ***Basil Al Suwaine***

Objectives

At the conclusion of these lectures the student will be able to:

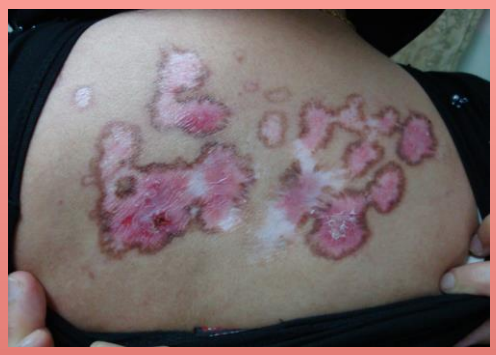
1. differentiate between the various types of Lupus
2. recognize how Lupus affects the various systems of the body
3. identify all of the current treatment options available for Lupus
4. recognize the psychosocial effects that Lupus has on the patient and their family
5. To learn how to diagnose and investigate dermatomyositis.
6. How to manage dermatomyositis.
7. To learn the presentation of morphea and systemic sclerosis and ways to manage them.
8. To recognize other diseases like Rheumatoid nodules and mixed CTD.
9. This lecture is not meant to be inclusive of all the information about these diseases but to highlight important aspects in their diagnosis and management.



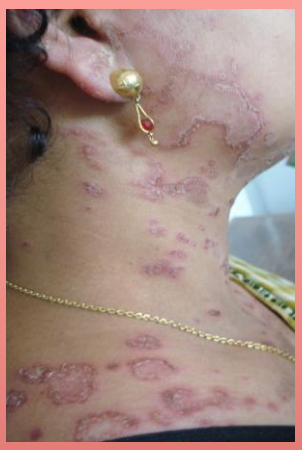
Lupus Erythematosus:

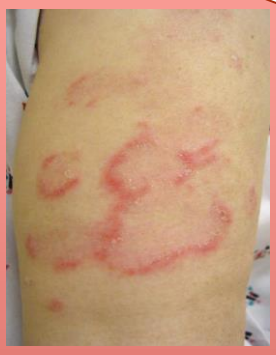
is as an autoimmune diseases associated with antibodies directed against components of cell nuclei. Lupus may affect any tissue, skin, kidneys, CNS, lungs and others.

Lupus Types:

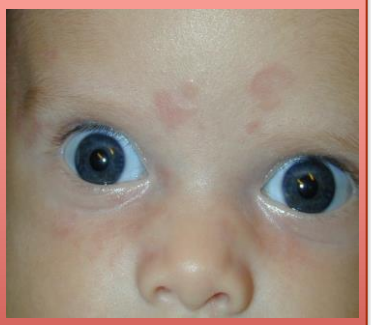


Discoid Lupus Discoid meaning "coin shape"
It is the commonest form of cutaneous lupus usually presents as red scaly patches or plaques that leave dyspigmentation and scarring mostly Hypopigmented or depigmented scars. It may be localized or widespread. Usually affects the cheeks, nose and ears, but sometimes involves the upper back, neck, and backs of hands.
Involvement of hair follicles will lead to scarring alopecia.
10% of DLE patients develop SLE. (so we frequently do ANA test)





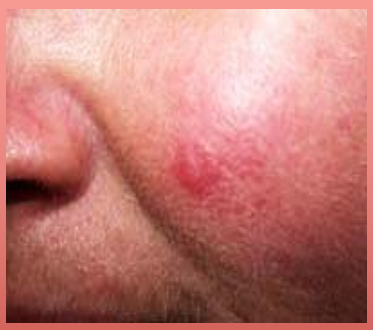
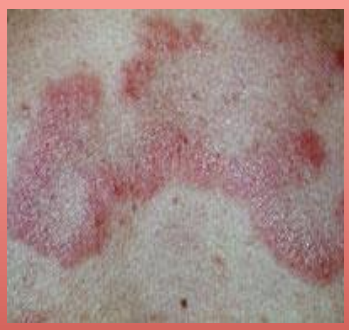
Subacute Lupus Erythematosus
Non-itchy dry rash appears on the upper back and chest, often following sun exposure.
Subacute LE does not scar.
Systemic involvement is not usually severe.
Annular or polycyclic (ring-shaped) or as papulosquamous (scaly patches and plaques)

Neonatal lupus erythematosus

Newborn babies born to mothers with subacute LE may develop annular rash, that resolve spontaneously.

The neonates could be at risk of **complete heart block.**

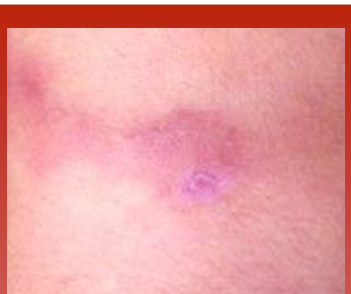
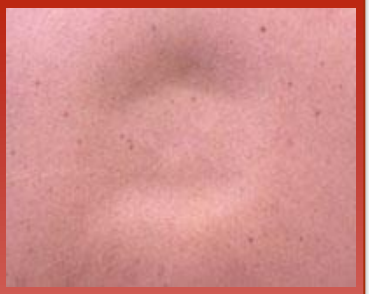



Lupus Tumidus (tumidus means swelling)

Dermal form of lupus.

The rash is characteristically photosensitive, so it affects sun-exposed sites.

Red, swollen, urticaria-like bumps and patches or swelling.

Lupus Profundus "Profundus Means deep"

Lupus affecting the fat underlying skin lupus panniculitis.

It may develop at any age, including children. The face is the most common area to be affected.

Inflammation of the fat results in firm deep nodules for some months.

The end result is deep scars on fat layer or lipodystrophy.

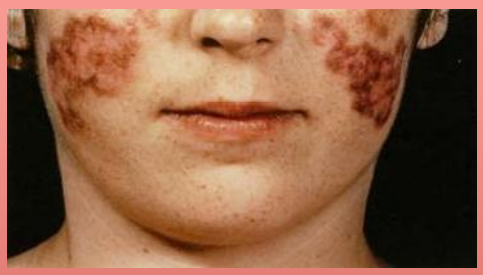
Chilblain Lupus Erythematosus "usually On cold area"

Itchy and/or tender red or purple bumps that usually come on from cold exposure but can sometimes be precipitated by sun exposure or smoking.

They are considered to be a form of skin vasculitis (blood vessel inflammation).

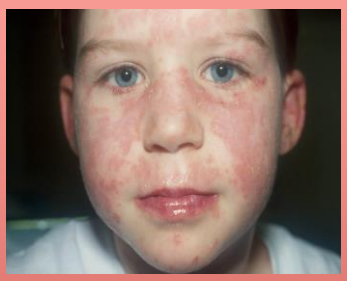
Usually they have no circulating antibodies.

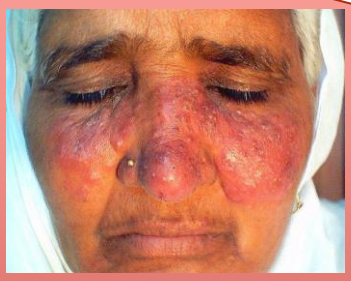
And the main treatment is to avoid precipitating factors.



Drug-Induced Lupus Erythematosus

Drug induced lupus does not usually affect the skin. The most frequent drugs are: **Hydralazine, Carbamazepine, Lithium, Phenytoin, Sulphonamides, Minocycline. (Very Important to know the drugs)**





Systemic Lupus Erythematosus

Only a few patients with cutaneous LE also have SLE.

The most common presentation is with a malar eruption or butterfly.

Other skin changes in SLE are photosensitivity, mouth ulcers, and diffuse hair loss.

SLE may also affect joints, kidneys, lungs, heart, liver, brain, blood vessels and blood cells.

Clinical Features:

Table 2. Clinical Features of SLE

System	Presentation
Constitutional	Fatigue, fever (in absence of infection), weight loss
Musculoskeletal	Arthritis, arthralgia, myositis
Skin	Butterfly rash, photosensitivitiy, mucous membrane lesion, alopecia, purpura, urticaria, vasculitis
Vascular	Raynaud's disease
Renal	Hematuria, proteinuria, renal casts, nephrotic syndrome
Gastrointestinal	Nausea, vomiting, abdominal pain
Pulmonary	Pleurisy, pulmonary hypertension
Cardiac	Pericarditis, endocarditis, myocarditis
Reticuloendothelial	Lymphadenopathy, splenomegaly, hepatomegaly
Hematologic	Anemia, thrombocytopenia, leukopenia
Neuropsychiatric	Psychosis, seizures, organic brain syndrome, transverse myelitis, cranial neuropathies, peripheral neuropathies

SLE: systemic lupus erythematosus.
Source: Reference 13.

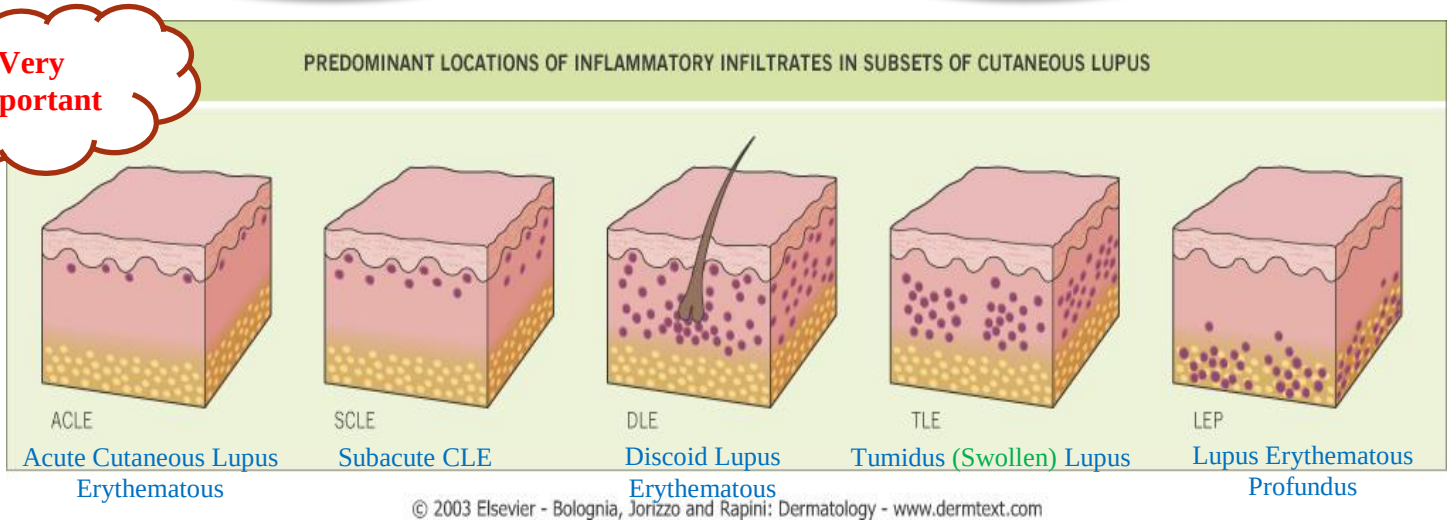
Investigations:

SLE is always with positive ANA.
Anti Ro/La antibodies, is nearly always present in patients with subacute LE.
Leucopenia tends to be more pronounced in patients with systemic LE.
Skin biopsy may be diagnostic especially in discoid lupus erythematosus.
Direct immunofluorescence tests may show positive antibody deposition along the basement membrane (lupus band test).

Treatment:

The aim of treatment for cutaneous LE is to alleviate symptoms and to prevent scarring.
Smoking cessation will help Raynaud's phenomena and chilblain lupus.
Sun protection.
Potent topical steroids, Intra lesional steroids. (but in The face
When don't use super potent steroid
Because it may cause thinning of the epidermis, folliculitis &telangiectasias).
Oral antimalarial drugs.
Oral steroids.
Methotrexate, azathioprin, mycophenolate mofetil, cyclosporine, cyclophosphamide, IVIG, and Rituximab.

Very Important



- **ACLE:** Very superficial and show only erythema so it doesn't cause scaring, Ex: malar rash.
- **SCLE:** Deeper than ACLE and manifest as scaly lesions and doesn't cause scaring.
- **DLE:** It affects the dermis mainly around the hair follicle so it causes significant scaring, Ex: Scaring Alopecia. **(Worst one)**
- **TLE:** It affects the dermis and it'll manifest as edematous skin lesion.
- **LEP:** It affects subcutaneous tissue, so clinically it appears as depression.

Dermatomyositis

An uncommon inflammatory disease affects adults between 40-60 (females mainly) and children 5-15.

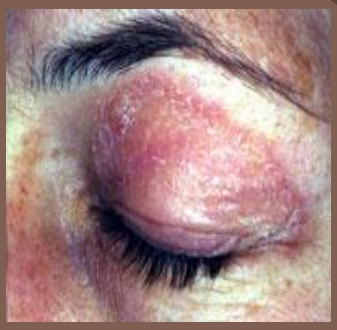
Skin changes: A violet-colored or dusky red rash on face and eyelids and on areas around nails, knuckles, elbows, knees, chest and back. The rash, which can be patchy with bluish-purple discolorations, is often the first sign of dermatomyositis.

Heliotrope rash: a violaceous -to-dusky erythematous rash with or without edema in a symmetrical distribution involving periorbital skin.

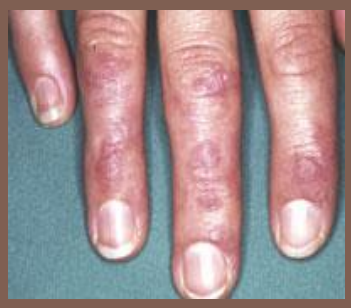
Muscle weakness: Progressive proximal muscle weakness involves the hips, thighs, shoulders, upper arms and neck. The weakness is symmetrical and more in the extensor muscles. (in progressive cases patients use wheelchair)

Other signs and symptoms include:
Photosensitivity
Raynaud's phenomenon
Dysphagia, gastrointestinal ulcers
Muscle pain or tenderness
Fatigue, fever and weight loss
Calcinosis cutis especially in children
Interstitial lung disease.

Periungual Telengectasia

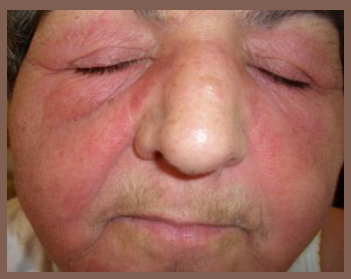


Heliotrope Rash

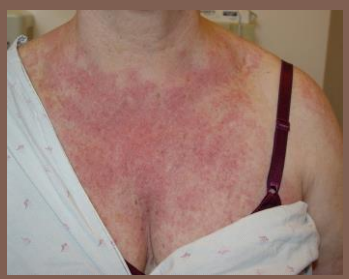


Gottron papules

Raynaud's phenomenon



Puffiens of cheek & eyelid



shawl sign rash

It can be associated with:
Other connective tissue diseases such as lupus, rheumatoid arthritis, scleroderma and Sjogren's syndrome.
Cancer, Especially in older patients, particularly of the cervix, lungs, pancreas, breasts, ovaries and gastrointestinal tract. Cancer could precede, coincide or follow the diagnosis of DM.

Investigations:

Magnetic resonance imaging (MRI).
Electromyography (EMG).
Muscle biopsy.
Blood tests: creatine kinase (CK) and aldolase. Increased CK and aldolase levels can indicate muscle damage and CK is useful to monitor the treatment of DM.
Autoantibodies
Skin biopsy is suggestive but not diagnostic that shows interface dermatitis.

Treatment:

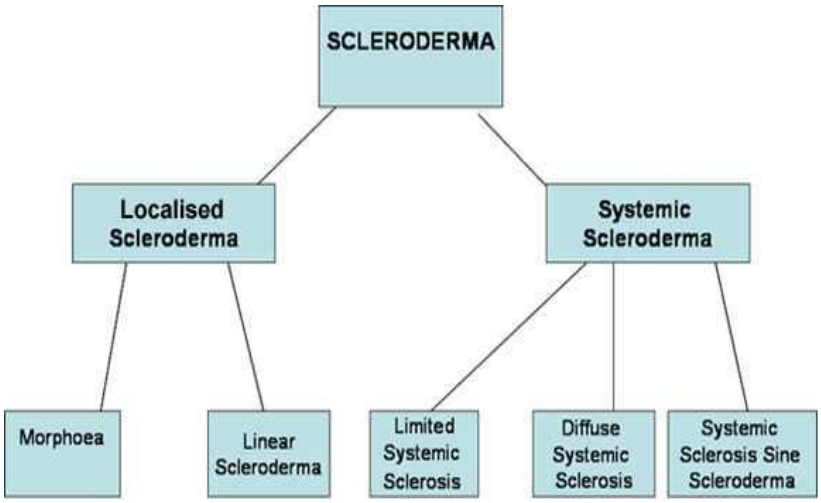
Oral steroids are the mainstay treatment.
Steroid sparing agents are: Methotrexate, azathioprin, mycophenolate mofetil, cyclosporine, cyclophosphamide, IVIG, and Rituximab.
Topical steroids and antimalarial medications are used to improve the cutaneous rashes.
Physiotherapy to improve strength and flexibility of the muscles.
Surgical excision or Co2 laser could be utilized to remove tender calcium deposits.

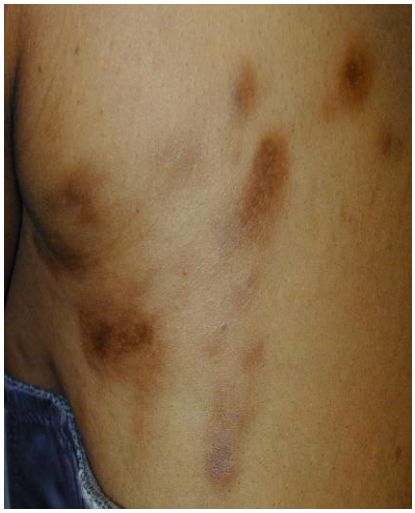
Scleroderma (Skin Sclerosis):

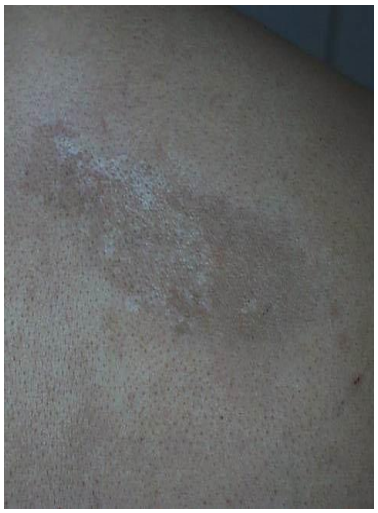
A group of rare diseases that involve the hardening and tightening of the skin and connective tissues. Scleroderma affects women more often than men and most commonly occurs between the ages of 30 and 50.

Morphea:

- a rare skin condition that causes oval reddish or purplish patches and plaques on the skin.
- Sometimes in linear distribution on face and extremities.
- It subsides on its own over time leaving dyspigmentation and scars.
- Confirmed by skin biopsy which usually shows thickening of collagen bundles and loss of skin appendages like sweat glands and hair follicles.
- No known cure.
- Treatment of morphea focuses on controlling signs and symptoms and slowing spread.
- Topical and intralesional steroids , phototherapy, systemic steroids, azathioprine, methotrexate, and cyclosporine might be used in severe cases.
- Physical therapy could be of help if the involvement is close to joints and cause contracture and difficulty movement.

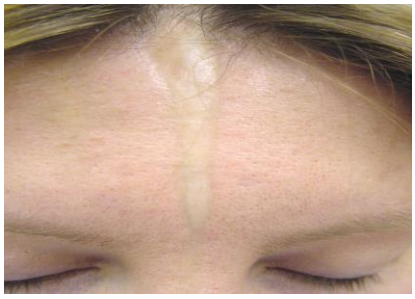


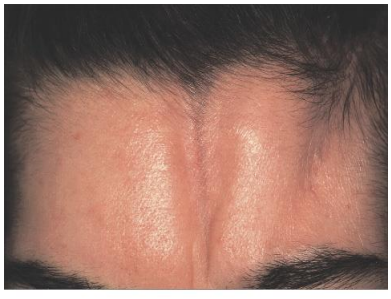


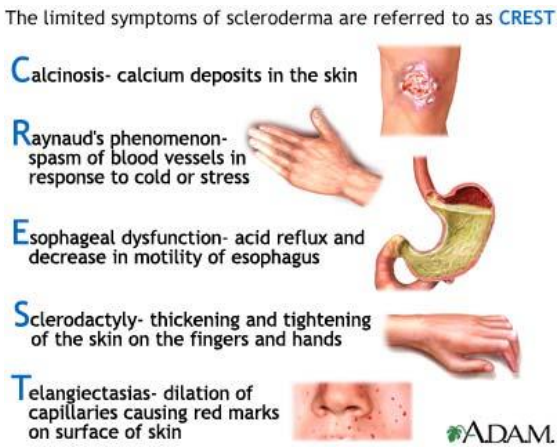


Linear Scleroderma (en coup de sabre):

- Linear lesions extend to length of arms or leg.
- Begin first decade of life.
- May also occur parasagitally down the forehead.



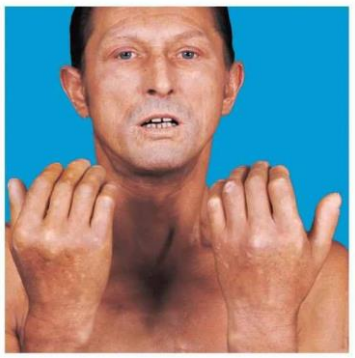


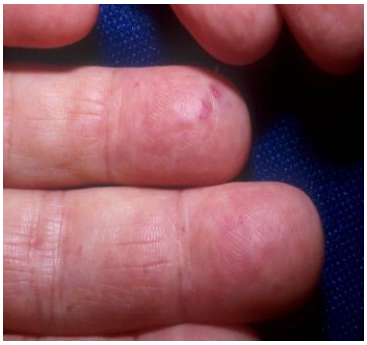


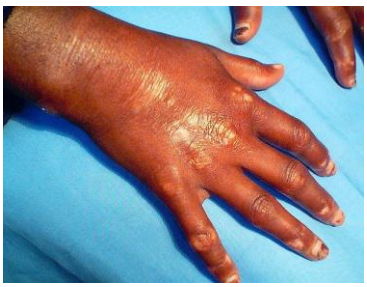
CREST Syndrome:

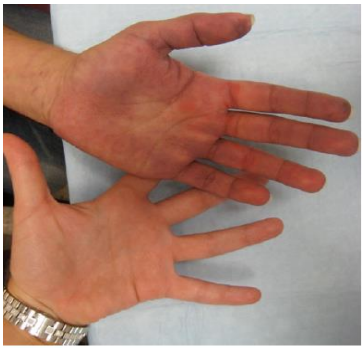
- Is a limited form of systemic sclerosis in which there is **C**alcinosis, **R**aynaud's phenomenon, **E**sophageal involvement, **S**clerodactyly and **T**elangiectasia.
- **Anticentromere antibodies** are characteristic for this syndrome.

Systemic Sclerosis:









- An autoimmune multisystem disease that results in fibrosis and vascular abnormalities in association with autoimmune changes.
- Usually starts between 30-40 years in women who are more affected and later in men.
- Pathophysiology: may involve some injury to the endothelial cells and this results in excessive activation of the dermal connective tissue cells, the fibroblasts.
- Usually presents with Raynaud's phenomena,
- Thickening of the skin of the fingers, then atrophy and sclerosis.
- The fingers become spindle-shaped (sclerodactyly) from resorption of the fingertips.
- Fragile nails become smaller with ragged cuticles
- The tight shiny skin may affect most parts of the body, including the face, resulting in loss of expression and difficulty opening the mouth properly.
- telangiectasia appear on the fingers, palms, face, lips, and chest.
- Ulcers may follow minor injuries over the joints, or on the tips of fingers and toes. Ulceration can lead to dry gangrene and eventual loss of the tips of the fingers
- Joint contractures. Patients will be bed ridden with time.
- Esophageal reflux and dysphagia.
- Lung and heart involvement may manifest as shortness of breath, high blood pressure, chest pain, pleurisy, pneumothorax, pericarditis arrhythmias, general heart enlargement and heart failure.
- Progressive kidney disease resulting in proteinuria, high blood pressure and eventually renal failure.

Investigations:

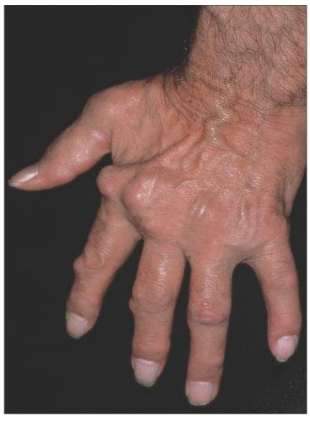
Diagnosis is made based on clinical features and presentation.
Skin biopsy will show skin atrophy with preservation of skin appendages.
ANA is usually **positive**.
Anti topoisomerase I (Scl 70) is characteristic for it especially in severe cases.

Treatment

symptomatic.
Raynaud's phenomena:
Stop smoking, keep hands warm and decrease trauma. calcium channel blockers, aspirin and vasodilating drugs including nifedipine and iloprost infusions.
Calcinosis cutis:
Nifedipine, surgical or laser excision.
Skin sclerosis:
physiotherapy, phototherapy.
GI: proton pump inhibitor, surgery for strictures.
Kidney: ACE inhibitors.
In severe cases:
immunosuppressant, D-Penicillamine might be used.

Rheumatoid Nodules:

- 20-30% of RA patients
- Subcutaneous nodules
- Found anywhere on the body
- Histologically shows dense foci of fibrinoid necrosis surrounded by histiocytes in palisaded arrangement



Mixed Connective Tissue Disease:

- Mixed features of scleroderma, SLE, and dermatomyositis
- Raynaud phenomenon, sausage-shaped fingers, and swelling of the dorsa of the hands that never becomes sclerodactyly are the most typical features
- Alopecia, facial erythema, periungual telangiectasia, and pigmentary disturbances.
- Painful dermal nodules may appear on the hands or elbows.
- Cutaneous ulceration due to subcutaneous dystrophic calcification

Very Important

Table 2: Useful Antibodies for CTD-ILD Assessment

Autoantibody	Associated CTD
High titer ANA (>1:320 titer)	Many
High titer RF (>60 IU/mL)	RA, Sjögren's disease, SLE
Anti-CCP	RA
Anti-centromere	Systemic sclerosis
Anti-nucleolar-ANA	Systemic sclerosis
Anti-Ro (SS-A)	Many
Anti-La (SS-B)	SLE, Sjögren's disease
Anti-Smith	SLE
Anti-ribonucleoprotein	SLE, MCTD
Anti-dsDNA	SLE
Anti-topoisomerase (Scl-70)	Systemic sclerosis
Anti-tRNA synthetase antibodies	Poly-/dermatomyositis (antisynthetase syndrome)
Anti-PM-Scl	Systemic slerosis/myositis overlap
Anti-Th/To	Systemic sclerosis
Anti-U3 ribonucleoprotein	Systemic sclerosis
ANCA panel	Systemic vasculitis

Summary (From FitzPatrick's book)

Lupus Erythematosus (LE)
ICD-9: 695.4 ◦ ICD-10: L93

- LE is the designation of a spectrum of disease patterns that are linked by distinct clinical findings and distinct patterns of cellular and humoral autoimmunity.
- LE occurs more commonly in women (male to female ratio 1:9).
- LE ranges from life-threatening manifestations of acute systemic LE (SLE) to the limited and exclusive skin involvement in chronic cutaneous LE (CCLE) (Fig. 14-32). More than 85% of patients with LE have skin lesions, which can be classified into LE specific and nonspecific.
- An abbreviated version of Gilliam classification of LE-specific skin lesions is given in Table 14-3.
- Acute cutaneous LE (ACLE) is practically always associated with SLE, subacute cutaneous LE (SCLE) in about 50%, and CCLE most often has only skin disease. However, CCLE lesions can occur in SLE.
- ACLE and SCLE are highly photosensitive.

Dermatomyositis ICD-9: 710.3 ◦ ICD-10: M33.0

- Dermatomyositis (DM) is a systemic disease belonging to the idiopathic inflammatory myopathies, a heterogeneous group of genetically determined autoimmune diseases targeting the skin and/or skeletal muscles.
- DM is characterized by violaceous (heliotrope) inflammatory changes +/– edema of the eyelids and periorbital area; erythema of the face, neck, and upper trunk; and flat-topped violaceous papules over the knuckles.
- It is associated with polymyositis, interstitial pneumonitis, and myocardial involvement.
- There is also a DM without myopathy (amyopathic DM) and polymyositis without skin involvement.
- Juvenile DM runs a different course and is associated with vasculitis and calcinosis.
- Adult-onset DM may be associated with internal malignancy.
- Prognosis is guarded.

Scleroderma ICD-9: 710.1 ◦ ICD-10: M34

- Scleroderma is a not so rare multisystem disorder characterized by inflammatory, vascular, and sclerotic changes of the skin and various internal organs, especially the lungs, heart, and GI tract.
- Limited systemic scleroderma (lSSc) (60%) and diffuse systemic scleroderma (dSSc) are recognized.
- Clinical features always present are skin sclerosis and Raynaud phenomenon.
- Considerable morbidity; high mortality of dSSc.
- *Synonyms:* Progressive systemic sclerosis, systemic sclerosis, systemic scleroderma.

QUESTIONS

1- A 50-year-old lady with dermatomyositis, you started her on systemic steroids.
To follow up this Patient, which one of the following can be used:
a. ANA
b. CBC and biochemistry
c. Creatine kinase
d. Muscle biopsy

2- A 20 years old male patients presented with dark red atrophic plaque his skin biopsy showed thickening of the dermal collagens and loss of the skin appendages.
What is the most likely diagnosis?
a. Morphea
b. Systemic sclerosis
c. Subacute lupus
d. Acute Lupus

3- A 40 year old woman presented with an eight months history of discoloration on the periorbital region and papules and plaques on the forearms and knuckles with thickened cuticles.
Which one of the following is the correct diagnosis:
a. Dermatomyositis
b. Discoid lupus erythematosus
c. Subacute lupus erythematosus
d. Systemic Sclerosis

4- Lady with scaly plaque with raised edges and hypopigmented depressed center.
What is the most proper investigation to reach the diagnosis
a-Anti smith.
b-Anti Ro.
c-Skin biopsy.
d-No need to investigate.

5- Patient came complaining of Muscle weakness with red edematous bilateral plaque over her eye lids.
Which one of the following signs she has?
a. Heliotrope.
b. Gottron papules.
c. Shawl sign.

1- C
2- A
3- A
4- C
5- A


Success is the sum of small efforts, repeated day in and day out.
Robert Collier