Connective Tissue Disorders

RHEUMATOLOGY.PHYSIO

A QUICK REFERENCE GUIDE TO:

Systemic Lupus Erythematosus
Sjögren's Syndrome
Polymyositis
And More...

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Contents

PLEASE REMEMBER – THIS GUIDE IS NOT A REPLACEMENT FOR CLINICAL REASONING, IF YOU ARE UNSURE GET ADVICE

Overview

In Rheumatology, a connective tissue disorder refers to a group of conditions that primarily affect the connective tissues of the body. Connective tissues provide structural support and integrity to various organs, joints, and other body structures.

Connective Tissue Disorders are characterized by abnormalities in the structure, function, or regulation of connective tissues. These disorders can involve multiple organ systems and may manifest in a variety of symptoms.

Some common Connective Tissue Disorders in Rheumatology include:

Systemic Lupus Erythematosus (SLE)
Sjögren's Syndrome
Undifferentiated Connective Tissue Disorder
Systemic Sclerosis (Scleroderma)
Polymyositis and Dermatomyositis
Hypermobility / Ehlers Danlos Syndrome(s)

Systemic Lupus
Erythematosus

Systemic Lupus Erythematosus (SLE) is a chronic autoinflammatory disease that commonly affects multiple organ systems in the body. Production of autoantibodies and chronic inflammation, leads to a wide range of symptoms and clinical manifestations. SLE predominantly affects female's and there is a higher incidence in people of African American decent.

The exact cause of SLE is not fully understood, but it is believed to involve a combination of genetic, hormonal, and environmental factors.

In SLE, the immune system begins to produce autoantibodies leading to the formation of immune complexes. These immune complexes can deposit in various tissues, promoting inflammation and tissue damage.

SLE can affect almost any organ system in the body, resulting in a wide range of clinical manifestations.

Systemic Lupus
Erythematosus

Presenting Features
Multiple joint pains
Myalgia
Fatigue

Comorbid/PMH
Other autoimmune condition(s)

Demographics
10:1 Female:Male
Twice as likely in African American women compared to Caucasian women
Peak onset age 15-44

Non-MSK Features
Butterfly Rash
Multiple miscarriages
Fatigue
Hair loss
Kidney Involvement

Family History
Lupus
Other autoimmune condition(s)

Imaging
Ultrasound if possible synovitis

Bloods
ANA +ve (95%)
Secondary care bloods anti-dsDNA antibodies, low complement, anti-sm antibodies

Sjögrens Syndrome

Sjögren's Syndrome is a chronic autoinflammatory disorder causing lymphocytic infiltration and destruction of the exocrine glands, primarily affecting the salivary and lacrimal glands. This results in the hallmark symptoms of dry mouth (xerostomia) and dry eyes (keratoconjunctivitis sicca).

Sjögren's Syndrome can be classified as **Primary** or **Secondary**. Primary Sjögren's syndrome occurs when the condition is not associated with any other autoimmune disease. Secondary Sjögren's syndrome occurs in the presence of another autoimmune disorder, such as rheumatoid arthritis or systemic lupus erythematosus.

Autoantibodies, such as anti-SSA (Ro) and anti-SSB (La) antibodies, are commonly detected in patients with Sjögren's syndrome and can aid in diagnosis.

The Schirmer's Test involves placing a small strip of filter paper inside the lower eyelid. The strip remains in place for a designated period of time, typically five minutes. The strip absorbs the tears produced by the eyes.

Sjögrens Syndrome

Presenting Features

Myalgia
Fatigue
Multiple joint pains

Comorbid/PMH

Other autoimmune condition(s)

Demographics

9:1 Female:Male
Peak onset age 50-60

Non-MSK Features

Dry eyes
Dry mouth
Vaginal Dryness
Fatigue

Family History

Sjogrens Syndrome
Other autoimmune condition(s)

Imaging

<5 mm in 5 minutes indicates a tear deficiency

Bloods

Anti-Ro/La antibodies positive

Undifferentiated CTDs

Undifferentiated Connective Tissue Disorder (UCTD) is a term used to describe a clinical condition with symptoms and laboratory findings that are suggestive of a Connective Tissue Disorder, but do not meet the specific diagnostic criteria for a specific diagnosis.

Patients with UCTD often present with a combination of symptoms commonly seen in CTDs, such as joint pain, swelling, and stiffness, along with systemic manifestations like fatigue, low-grade fever and skin rashes. The symptoms are often milder than those with clear SLE for example.

The presence of **specific autoantibodies** can provide supporting evidence for UCTD. Commonly observed autoantibodies include antinuclear antibodies (ANA) and anti-SSA (Ro) and anti-SSB (La) antibodies. However, the absence of these autoantibodies does not exclude a diagnosis of UCTD.

Undifferentiated CTDs

Presenting Features

Myalgia
Fatigue
Multiple joint pains
Milder Symptoms

Comorbid/PMH

Other autoimmune condition(s)

Demographics

9:1 Female:Male
Peak onset age 32-44
72% Caucasian

Non-MSK Features

Dry eyes
Dry mouth
Fatigue
Butterfly Rash
Organ involvement
Neurological Symptoms

Family History

Other autoimmune condition(s)

Imaging

Ultrasound if possible synovitis

Bloods

One Auto-antibody positive ANA/Anti-Ro/Anti-La

Systemic Sclerosis

Scleroderma

Systemic Sclerosis (Scleroderma), is a chronic autoinflammatory Connective Tissue Disorder causing excessive fibrosis (thickened and hardened) of the skin and internal organs. The skin symptoms usually affects the palms of the hands effectively causing flexion contractures.

Systemic Sclerosis is relatively rare, with an estimated prevalence of 10 to 20 cases per million. Female diagnostic predominance is 3:1. Onset is typically between ages 30 and 50, but it can occur at any age.

The inflammatory process causes damage to small blood vessels, triggers excessive collagen and extracellular matrix proteins leading to fibrosis of the skin and some internal organs.

In addition to the skin thickening Raynaud's phenomenon is almost always present, Gastrointestinal and Pulmonary involvement is common. Musculoskeletal manifestations of inflammatory arthritis mainly affect the hands and fingers. Rarely there is also Cardiac and Renal involvement.

Systemic Sclerosis

Scleroderma

Presenting Features

Skin Fibrosis (particularly palms)

Raynaud's Phenomenon

Inflammatory Arthritis (hands)

Comorbid/PMH

Other autoimmune condition(s)

Demographics

3:1 Female:Male

Peak onset age 30-50

Non-MSK Features

Reflux

Dysphagia

Impaired Lung Function

Pulmonary Hypertension

Bloods

ANA

Rheumatoid Factor

ESR & CRP

Creatine Kinase (CK)

Liver Function Tests

Kidney Function

Imaging

Ultrasound if possible synovitis

Doppler (Raynaud's Phenomenon

Myositis

Polymyositis & Dermatomyositis

Polymyositis and Dermatomyositis are autoinflammatory muscle diseases in the category of Idiopathic Inflammatory Myopathies. There are similarities but also some distinct clinical features.

Polymyositis primarily involves the proximal muscles of the limbs and then the trunk causing progressive symmetrical weakness and discomfort.

Dermatomyositis presents with a similar pattern to Polymyositis but with a characteristic skin rash. The rash typically appears on the face, eyelids, neck, chest, elbows, knees, or knuckles. It can be erythematous, rash-like, or present as a heliotrope rash (reddish-purple discoloration around the eyes).

Systemic symptoms are almost always present, commonly these are fatigue, malaise, fever and weight loss.

Blood testing reveals raised Creatine Kinase, Inflammatory Markers and auto-antibodies (anti-jo, anti-sRP)

Myositis

Polymyositis & Dermatomyositis

Presenting Features

Progressive Proximal Muscle Weakness

Comorbid/PMH

Other autoimmune condition(s)

Demographics

Polymyositis

1:1 Female:Male

Peak onset age 30-60

Dermatomyositis

2:1 Female:Male

Peak age onset 5-15 & 45-65

Non-MSK Features

Erythematous Rash

Heliotrope Rash

Fatigue

Weightloss

Malaise

Fever

Bloods

Creatinie Kinase (CK) >500

ESR & CRP

Anti-Jo/Anti-SRP

Other Investigation

Muscle Biopsy

MRI or Ullrasound to view muscle inflammation

Lupus

CLINICAL SCENARIO

Referral

Mary is a 30 year old female with diffuse aching all around her body, she doesn't take any regular medications.

Further Information

Mary has had diffuse aching all around her body for approximately 6 weeks. The intensity and location of the aching is variable and seems related to how tired she is. She has always been a poor sleeper but in the last 6 weeks she has had significantly worse fatigue. She generally feels unwell and run down but denies any specific symptoms like nausea, dizziness or pyrexia.

Mary reports she has had 3 miscarriages over the last 7 years. She and her husband are continuing to try and have a baby. She has also had a rash across her face at times over the last 6 weeks which has not been itchy or sore. It is not visually present in clinic today.

Clinical Reasoning Activity

From the case presentation note down the indicators that Anna could have Systemic Lupus Erythematosus.

Sjögrens Syndrome

CLINICAL SCENARIO

Referral

Anna is a 40 year old female with diffuse muscle aching in her legs. She doesn't take any prescribed medication.

Further Information

Anna has had diffuse muscle aching in her legs for approximately 6 months. There was no triggering onset and no change in habits prior. More recently over the last 4-6 weeks she has felt very tired all of the time. She reports sleeping well but never feels refreshed.

Anna reports very dry eyes. She uses false tears regularly through the day and also has a very dry mouth. She denies any change in vaginal dryness. She carries a bottle of water everywhere with her and drinks from it regularly during the appointment.

Clinical Reasoning Activity

From the case presentation note down the indicators that Anna could have Sjogren's Syndrome

More At A Glance

Rheumatology
Spinal Masqueraders
The Hip
The Hand
Lumbar Radicular Syndromes
The Knee
The Foot

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