Rheumatologic Diseases: Clinical Features, Diagnosis, and Treatment

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Outline

- RA,
- SLE,
- Sjögren's disase
- OA,
- Gout,
- Fibromyalgia

Rheumatoid Arthritis - Introduction & Epidemiology

- Chronic autoimmune inflammatory arthritis, synovial joints.
- Global prevalence ~1%; onset 35–50 yrs.
- Female:Male ratio ~3:1.
- Genetic predisposition: 2–3x risk in first-degree relatives.
- Environmental triggers: smoking, infections.

Rheumatoid Arthritis - Clinical Manifestations

- Symmetric polyarthritis: MCP, PIP, wrists, MTP.
- Morning stiffness >30 min, improves with activity.
- Fatigue, malaise, low-grade fever, weight loss.
- Progressive → joint destruction, deformity.
- Cervical spine involvement possible.

Rheumatoid Arthritis
Organ Involvement

- Skin: rheumatoid nodules.
- Lungs: ILD, pleuritis, nodules.
- Heart: pericarditis, ↑atherosclerosis.
- Eyes: scleritis, episcleritis, keratoconjunctivitis sicca.
- Hematologic: anemia, Felty's syndrome.

Rheumatoid Arthritis - Diagnosis

- Persistent symmetric polyarthritis.
- Serology: RF, Anti-CCP (specific).
- ESR, CRP correlate with activity.
- Synovial fluid: inflammatory, neutrophil predominance.
- Imaging: X-ray (narrowing, erosions), MRI/US for early disease.

Rheumatoid Arthritis - Treatment

- Treat-to-target: remission/low activity (DAS28, CDAI).
- Methotrexate = first-line DMARD.
- Other DMARDs: leflunomide, sulfasalazine, hydroxychloroquine.
- Biologics: TNF, IL-6 inhibitors, abatacept, rituximab.
- NSAIDs, steroids for symptoms; non-drug: PT, exercise.

SLE - Introduction & Epidemiology

- Chronic multisystem autoimmune disease.
- Predominantly women (9:1), onset 15–45 yrs.
- Prevalence: 20–150/100,000, varies by ethnicity.
- Higher incidence in African American, Hispanic, Asian.
- Genetic, environmental, hormonal triggers.

SLE - Clinical Manifestations

- Constitutional: fatigue, fever, weight loss.
- Musculoskeletal: non-erosive arthritis/arthralgia.
- Skin: malar rash, photosensitivity, discoid lesions.
- Renal: lupus nephritis.
- Neuropsychiatric: seizures, psychosis.

SLE - Organ Involvement

- Kidneys: lupus nephritis (class I–VI).
- Skin: malar rash, ulcers, alopecia.
- CNS: seizures, psychosis, stroke.
- Cardiac: pericarditis, Libman-Sacks endocarditis.
- Pulmonary: pleuritis, ILD, PH; Hematologic: anemia, cytopenia.

SLE - Diagnosis

- Clinical + immunologic criteria (EULAR/ACR 2019).
- ANA: sensitive (>95%), not specific.
- Specific: anti-dsDNA, anti-Smith, antiphospholipid.
- Low complements (C3, C4) in active disease.
- Urinalysis/biopsy for nephritis; exclude mimics.

SLE - Treatment

- Hydroxychloroquine: cornerstone therapy.
- Glucocorticoids for flares; taper to lowest dose.
- Immunosuppressives: azathioprine, mycophenolate, cyclophosphamide.
- Biologics:Rituximab, belimumab, anifrolumab.
- Preventive: CV risk, osteoporosis, vaccination, sun protection.

Sjögren's - Introduction & Epidemiology

- Chronic autoimmune exocrinopathy of salivary/lacrimal glands.
- Prevalence: 0.1–0.6%.
- Female predominance 9:1, onset 40–60 yrs.
- Primary or secondary (RA, SLE, systemic sclerosis).
- Pathogenesis: lymphocytic gland infiltration.

Sjögren's - Clinical Manifestations

- Sicca: dry eyes, dry mouth.
- Eye: burning, foreign-body sensation, photophobia.
- Oral: dysphagia, caries, parotid swelling.
- Fatigue, arthralgia common.
- Chronic course, variable severity.

Sjögren's - Organ Involvement

- Joints: non-erosive arthritis.
- Lungs: ILD, bronchiectasis.
- Kidneys: interstitial nephritis, RTA.
- Skin: xeroderma, vasculitis, Raynaud's.
- ↑Risk of MALT lymphoma.

Sjögren's - Diagnosis

- Clinical: sicca symptoms + systemic features.
- Autoantibodies: anti-Ro/SSA, anti-La/SSB, ANA, RF.
- Schirmer's test, ocular staining, salivary flow.
- Minor salivary gland biopsy (gold standard).
- 2016 ACR/EULAR classification criteria.

Sjögren's - Treatment

- Symptomatic: artificial tears, saliva substitutes.
- Systemic: hydroxychloroquine, steroids, immunosuppressants.
- Severe: rituximab, other biologics.
- Avoid anticholinergics.
- Regular lymphoma surveillance.

Osteoarthritis
Introduction & Epidemiology

- Most common arthritis; degenerative joint disease.
- >30% of people >65 yrs affected.
- Women more after menopause.
- Risk factors: age, obesity, injury, genetics, stress.
- Common joints: knee, hip, spine, hand.

Osteoarthritis - Clinical Manifestations

- Pain worse with activity, relieved by rest.
- Stiffness <30 min after inactivity.
- Crepitus, bony enlargement, ↓ROM.
- Hands: Heberden's (DIP), Bouchard's (PIP).
- Usually asymmetric, oligoarticular.

Osteoarthritis - Organ Involvement

- Localized musculoskeletal disease.
- Periarticular: osteophytes, ligament laxity, weakness.
- Secondary: immobility → obesity, metabolic syndrome.
- Rare: deformity, cervical spinal compression.

Osteoarthritis - Diagnosis

- Clinical: pain, stiffness, functional loss.
- X-ray: joint space narrowing, osteophytes, sclerosis, cysts.
- MRI: early cartilage changes, marrow lesions.
- Labs: normal (ESR, CRP).
- Exclude RA, gout, CPPD.

Osteoarthritis - Treatment

- Lifestyle: weight loss, exercise, PT/OT.
- Drugs: acetaminophen, NSAIDs, duloxetine.
- Intra-articular steroids for flares.
- Viscosupplementation in select patients.
- Joint replacement if severe.

Gout - Introduction & Epidemiology

- Metabolic disorder: urate crystal deposition.
- Prevalence ~4% adults; more in men.
- Risk: purine diet, alcohol, obesity, CKD, diuretics.
- Strong genetic component.
- Classic site: 1st MTP (podagra).

Gout - Clinical Manifestations

- Acute gouty arthritis: severe pain, swelling, erythema.
- Often nocturnal; rapid onset.
- Other sites: ankle, knee, wrist, elbow.
- Chronic: tophi, deformity, polyarthritis.
- Systemic: fever, malaise in flares.

Gout - Organ Involvement

- Joints: recurrent arthritis, tophi.
- Kidneys: stones, chronic urate nephropathy.
- Skin: tophi over joints, ears, tendons.
- Systemic inflammation: ↑CV risk.
- Can mimic septic arthritis.

Gout - Diagnosis

- Gold standard: urate crystals in synovial fluid.
- Serum uric acid often ↑, but can be normal in flare.
- X-ray: erosions with overhanging edges, tophi.
- US: double contour sign.
- Differentiate CPPD, septic arthritis, RA.

Gout - Treatment

- Acute: NSAIDs, colchicine, steroids.
- Prophylaxis: colchicine/NSAIDs during urate-lowering.
- Urate-lowering: allopurinol, febuxostat (goal <6 mg/dL).
- Probenecid in select patients.
- Lifestyle: diet, alcohol moderation, weight control.

Fibromyalgia
Introduction & Epidemiology

- Chronic widespread pain, central sensitization.
- Prevalence 2–4%.
- Female:Male ~9:1.
- Associated: depression, anxiety, IBS.
- Pathophysiology: altered pain processing.

Fibromyalgia - Clinical Manifestations

- Widespread pain ≥3 months.
- Fatigue, non-restorative sleep, cognitive dysfunction.
- Headaches, paresthesias, IBS.
- Tender points (not required in new criteria).
- Symptoms fluctuate with stress, sleep, weather.

Fibromyalgia - Organ Involvement

- Primarily CNS: central sensitization.
- No synovitis, no systemic organ damage.
- Autonomic: palpitations, dizziness, OI.
- Psychiatric comorbidities common.
- Significant disability but no ↑mortality.

Fibromyalgia - Diagnosis

- Clinical: WPI + symptom severity (ACR 2010/2016).
- Exclude mimics: hypothyroid, RA, SLE, PMR.
- Labs: normal ESR, CRP.
- Imaging not diagnostic.
- Often delayed diagnosis (2–5 yrs).

Fibromyalgia - Treatment

- Multidisciplinary: education, exercise, CBT.
- Drugs: duloxetine, milnacipran, pregabalin.
- NSAIDs, acetaminophen often ineffective.
- Sleep hygiene, stress management.
- Avoid chronic opioids.
- Focus: symptom relief, function.