Autoimmune and Autoinflammatory Diseases – Complete Overview

1 What Are They?

Autoimmune diseases occur when the adaptive immune system mistakenly attacks the bodys own cells using autoantibodies or autoreactive T-cells. Autoinflammatory diseases (periodic fever syndromes) occur due to malfunctions in the innate immune system, leading to recurrent inflammation without infection or autoantibodies.

2 Why Do They Occur?

- Genetic predisposition (HLA genes, immune-related mutations).
- Environmental triggers (infections, pollution, stress, diet, smoking).
- Hormonal factors (many autoimmune diseases are linked to estrogen).
- Epigenetic changes that misprogram immune cells.

3 Who Gets Them?

- **Gender**: Around 80% of autoimmune disease patients are women.
- **Age**: Most start between 2060 years. Some occur in childhood (Type 1 Diabetes, Juvenile Arthritis, PFAPA).
- **Geography**: More common in developed countries such as the US, Canada, and Europe. Some diseases are more frequent in specific ethnic groups (e.g., Familial Mediterranean Fever in Middle Eastern and Mediterranean populations).

4 Main Autoimmune Diseases

4.1 Rheumatoid Arthritis (RA)

- Immune attack on joints.
- Affects $\sim$1% of adults, mostly women (age 4060).
- **Symptoms**: Joint pain, swelling, stiffness, deformity.
- **Treatment**: NSAIDs, steroids, DMARDs, biologics.

4.2 Systemic Lupus Erythematosus (SLE)

- Immune attack on multiple organs.
- Female-to-male ratio ∼9:1.
- More common in US and France, less in Japan/Iceland.
- **Symptoms**: Skin rash, arthritis, kidney inflammation, fatigue.
- **Treatment**: Steroids, hydroxychloroquine, immunosuppressants, biologics.

4.3 Sjögrens Syndrome

- Immune attack on tear and salivary glands.
- 90% of patients are women, age 4060.
- **Symptoms**: Dry eyes, dry mouth, joint pain.
- **Treatment**: Symptomatic care, immunosuppressants if needed.

4.4 Hashimotos Thyroiditis and Graves Disease

- Immune attack on thyroid gland (hypothyroidism in Hashimotos, hyperthyroidism in Graves).
- Hashimotos prevalence ∼5%, Graves ∼0.5%.
- **Treatment**: Hormone replacement (Hashimotos), anti-thyroid drugs or surgery (Graves).

4.5 Type 1 Diabetes

- Immune destruction of pancreatic beta-cells.
- Usually starts in childhood/adolescence.
- **Treatment**: Insulin therapy, glucose monitoring.

4.6 Multiple Sclerosis (MS)

- Immune attack on central nervous system myelin.
- Onset 2040 years, more common in women.
- **Treatment**: Steroids, disease-modifying therapies, symptom management.

4.7 Inflammatory Bowel Disease (Crohns Disease, Ulcerative Colitis)

- Immune-mediated inflammation of digestive tract.
- More common in Western countries.
- **Symptoms**: Abdominal pain, diarrhea, bleeding.
- **Treatment**: Anti-inflammatory drugs, biologics, sometimes surgery.

4.8 Celiac Disease

- Immune reaction to gluten damaging intestinal lining.
- Prevalence $\sim$1% worldwide.
- **Treatment**: Strict gluten-free diet.

5 Autoinflammatory (Periodic Fever) Syndromes

5.1 Familial Mediterranean Fever (FMF)

- Genetic disease (MEFV mutation).
- Episodes of fever, abdominal/chest/joint pain.
- Common in Middle Eastern, North African, and Mediterranean populations.
- **Treatment**: Colchicine, biologics if resistant.

5.2 PFAPA Syndrome

- Stands for Periodic Fever, Aphthous Stomatitis, Pharyngitis, Adenitis.
- Onset age 25 years, usually resolves by age 10.
- **Symptoms**: Fever, sore throat, swollen lymph nodes, mouth ulcers.
- **Treatment**: Corticosteroids during attacks.

5.3 TRAPS (TNF Receptor Associated Periodic Syndrome)

- Autosomal dominant mutation.
- **Symptoms**: Recurrent fever, rash, muscle pain.
- **Treatment**: Biologics (IL-1 blockers, anti-TNF agents).

5.4 MKD (Hyper-IgD Syndrome)

- Defect in mevalonate kinase enzyme.
- Onset before 1 year of age.
- **Symptoms**: Fever, rash, swollen lymph nodes.
- **Treatment**: NSAIDs, steroids, biologics.

5.5 CAPS (Cryopyrin-Associated Periodic Syndromes)

- Includes Muckle-Wells Syndrome.
- **Symptoms**: Fever, hives-like rash, joint pain, hearing loss.
- **Treatment**: IL-1 inhibitors (anakinra, canakinumab).

6 Summary Table

Disease	**Target System**	**Onset Age**	**Gender Ratio (F:M)**	**Prevalence / Region**	**Treatment**
Rheumatoid Arthritis	Joints	4060	23:1 female	1% adults worldwide	DMARDs, steroids
Lupus (SLE)	Multiple organs	315 / 4564	9:1 female	High in US/France, low in Japan	Steroids, im pressants, b
Sjögrens Syndrome	Glands	4060	10:1 female	0.21.2%	Symptomati munosuppre
Hashimotos/Graves	Thyroid	3050	810:1 female	Common in women	Hormones thyroid ther
Type 1 Diabetes	Pancreas	Childhood	1:1	1% worldwide	Insulin
Multiple Sclerosis	CNS	2040	23:1 female	High in Europe, North America	Disease-moc therapy
IBD (Crohns/UC)	Gut	Teens30s	Slightly female	More common in West	Anti-inflamm drugs, biolo
Celiac Disease	Gut lining	Any age	12:1 female	1% globally	Gluten-free
FMF	Innate immune	Childhood	Equal or slight male	Mediterranean, Middle East	Colchicine,
PFAPA	Innate immune	25 years	1:1	Rare, self-limiting	Corticostero
TRAPS / MKD / CAPS	Innate immune	Childhood/adult	Variable	Extremely rare	IL-1 inhibit TNF

7 Key Points

- Autoimmune diseases involve adaptive immunity and autoantibodies.

- Autoinflammatory diseases involve innate immunity and recurrent fevers.
- Women are more affected by autoimmune diseases.
- Treatments include immunosuppressants, biologics, and symptomatic care.
- No universal cure yet; early diagnosis improves outcomes.