Autoimmune Disorders

What are autoimmune diseases?
There are more than 80 known types of autoimmune disease that affect various body systems. These conditions occur when a person’s immune system produces autoantibodies that attack their own cells as foreign (immune system reactions directed against intrinsic body components). In addition, the regulatory T cells, are not responding effectively. This response is a hypersensitivity reaction similar to the response in allergic conditions.

How common are autoimmune diseases and who is at risk?
Autoimmune diseases are common, affecting more than 23.5 million Americans. They affect from 15-50/100,000 people depending on the disease and are a leading cause of death and disability globally. Age of onset is between 15-40 years old, with peak onset in the 20s. Women are more prone to these conditions (M<F, 1:6-10). Higher risk groups include: those with a family history (and may inherit a relevant gene), those exposed to certain environmental factors (i.e. ultraviolet light or radiation, drugs, chemicals, bacterial and viral infections), those of a certain race or ethnicity (e.g.s. Type 1 diabetes is more common in Caucasians and Lupus may be more severe in African Americans and Hispanics).

Some autoimmune diseases and common symptoms:
Many autoimmune disease share hallmark symptoms: such as fatigue, malaise, dizziness, and a low-grade fever. Symptoms can wax and wane, go into remission or flare up.

An autoimmune disorder may result in:
- The destruction of one or more types of body tissue
- Abnormal growth of an organ
- Changes in organ function

Clinical presentation may include:
- Constitutional – fever, anorexia, weight loss
- Musculoskeletal – arthralgias, arthritis, synovitis, myopathy
- Dermatologic – skin rashes, Raynaud phenomenon, photosensitivity
- Cardiopulmonary – pleuritis, pericarditis, fibrosis, chest pain
- Renal – proteinuria, glomerulonephritis
- Otorhinolaryngologic – sicca syndrome, oral ulcers
- Gastrointestinal – gastroesophageal reflux disease
• Neurologic – seizures, encephalopathy
• Hematologic – cytopenias (involving neutrophils, erythrocytes and platelets)

Some common autoimmune diseases and related symptoms:

Alopecia areata
The immune system attacks hair follicles and can cause patchy hair loss on the scalp, face, or other areas of the body.

Antiphospholipid antibody syndrome (aPL)
A disease that causes problems in the inner lining of blood vessels resulting in blood clots in arteries or veins.
• Blood clots in veins or arteries
• Multiple miscarriages
• Lacy, net-like red rash on the wrists and knees

Autoimmune hepatitis
The immune system attacks and destroys the hepatic cells. This can lead to scarring and hardening of the liver, and possibly liver failure.
• Fatigue
• Hepatomegaly
• Jaundice and/or icterus
• Pruritis
• Joint pain
• Abdominal pain

Celiac disease
When gluten is eaten, the immune system responds by damaging the lining of the small intestines.
• Abdominal bloating and pain
• Diarrhea or constipation
• Weight loss or weight gain
• Fatigue
• Amenorrhea
• Pruritic dermatitis
• Infertility or miscarriages

Diabetes type 1 (IDDM)
A disease in which the immune system attacks pancreatic beta cells, resulting in a decrease or lack of insulin production. Hyperglycemia can damage the eyes, kidneys, nerves, gums and teeth, and cardiovascular system.
• Polydipsia
• Polyuria

• Feeling very hungry or tired
• Unexplained weight loss
• Slow healing of skin
• Dry, pruritic skin
• Paresthesia
• Blurry vision

*Graves' disease* (overactive thyroid)
• Insomnia
• Irritability
• Weight loss
• Heat sensitivity
• Diaphoresis
• Fine brittle hair
• Muscle weakness
• Light menstrual periods
• Bulging eyes
• Tremors
• Tachycardia

*Guillain-Barre syndrome*
The immune system attacks the nerves that connect the brain and spinal cord with the rest of your body. Nerve damage results.
• Weakness or tingling feeling in the legs that might spread to the upper body
• Paralysis in severe cases

Symptoms often progress relatively quickly, over a period of days or weeks, and often occur bilaterally.

*Hashimoto's disease* (underactive thyroid)
• Fatigue
• Weakness
• Weight gain
• Sensitivity to cold
• Muscle aches and stiff joints
• Facial swelling
• Constipation
• Amenorrhea or dysmenorrhea

*Hemolytic anemia*
A condition in which the immune system destroys the red blood cells. As a result, hypoxemia occurs which causes the heart to work harder to move oxygen-rich blood throughout the body.
• Fatigue
• Shortness of breath
• Dizziness
• Headache
• Cold hands or feet
• Paleness
• Jaundice and/or icterus
• Cardiovascular problems, including heart failure

*Idiopathic thrombocytopenic purpura (ITP)*
A disease in which the immune system destroys platelets, which are needed for blood to clot.
• Dysmenorrhea or menorrhagia
• Purpuric rash
• Easy bruising
• Epistaxis or bleeding in the mouth

*Inflammatory bowel disease (IBD)*
A disease that causes chronic inflammation of the digestive tract. Crohn's disease and ulcerative colitis are the most common forms of IBD.
• Abdominal pain
• Diarrhea, which may be bloody
• Some people also have:
• Rectal bleeding
• Fever
• Weight loss
• Fatigue
• Mouth ulcers (in Crohn's disease)
• Painful or difficult bowel movements (in ulcerative colitis)

*Inflammatory myopathies*
A group of diseases that involve muscle inflammation and muscle weakness. **Polymyositis and dermatomyositis** are 2 types more common in women than men.

Slow but progressive muscle weakness beginning in the muscles closest to the trunk of the body. Polymyositis affects muscles involved with making movement on both sides of the body. With dermatomyositis, a skin rash comes before or at the same time as muscle weakness.

May also have:
• Fatigue after standing or ambulation
• Loss of balance
• Dysphagia or dyspnea

*Multiple sclerosis (MS)*
A disease in which the immune system attacks myelin sheath. The damage affects the brain and spinal cord.
• Weakness and trouble with coordination, balance, speaking, and walking
• Paralysis
• Tremors
• Paresthesia in arms, legs, hands, and feet
• Symptoms vary

**Myasthenia gravis (MG)**
A disease in which the immune system attacks the nerves and muscles throughout the body.
• Diplopia, trouble keeping a steady gaze, and ptosis
• Dysphagia, with frequent gagging or choking
• Weakness or paralysis
• Muscles that work better after rest
• Drooping head
• Trouble climbing stairs or lifting things
• Trouble talking or aphasia

**Primary biliary cirrhosis**
The immune system slowly destroys the liver’s bile ducts. Bile builds up in the liver and causes damage. The damage causes the liver to harden and scar, and eventually stop working.
• Fatigue
• Pruritis
• Dry eyes and mouth
• Jaundice and/or icterus

**Psoriasis**
• Thick scaly red patches on skin usually appearing on the head, elbows, and knees
• Itching and pain, which can make it hard to sleep and ambulate

May also have:
• Psoriatic arthritis that often affects the joints and the ends of the fingers and toes. Back pain can occur if the spine is involved.

**Rheumatoid arthritis**
• Painful, stiff, swollen, and deformed joints
• Reduced range of motion and function
• Fatigue
• Fever
• Weight loss
• Eye inflammation
• Lung disease
• Nodules
• Anemia

**Scleroderma**
• Abnormal growth of connective tissue in the skin and blood vessels.
• Fingers and toes that turn white, red, or blue in response to heat and cold
• Pain, stiffness, and swelling of fingers and joints
• Lichenification
• Skin that looks shiny on the hands and forearm
• Tight and mask-like facial skin
• Sores on the fingers or toes
• Dysphagia
• Weight loss
• Diarrhea or constipation
• Shortness of breath

**Sjögren’s syndrome**
A disease in which the immune system targets the glands that make moisture, such as tears and saliva.

- Dry eyes and pruritic eyes, cloudy eyes
- Dryness of the mouth, which can cause sores
- Dysphagia
- Loss of sense of taste (ageusia)
- Severe dental cavities
- Hoarse voice
- Fatigue
- Joint swelling or pain
- Swollen glands

**Systemic lupus erythematosus (SLE)**
A disease that can damage the joints, skin, kidneys, heart, lungs, and other parts of the body.

- Fever
- Weight loss
- Alopecia
- Mouth sores
- Fatigue
- "Butterfly" rash across the nose and cheeks and on other areas of the body
- Painful or swollen joints and myalgia
- Sensitivity to the sun
- Chest pain
- Headache, dizziness, seizure, memory problems, or change in behavior

**Vitiligo**
The immune system destroys the cells that gives skin its color. It also can affect the tissue inside the mouth and nose.

- White patches on areas exposed to the sun, or on armpits, genitals, and rectum
- Hair turns gray early
- Loss of color inside your mouth

For a more thorough list of autoimmune diseases and related symptoms, refer to the American Autoimmune Related Disease Association website - [http://www.aarda.org/research_display.php?ID=47](http://www.aarda.org/research_display.php?ID=47)
Assessment

- A complete personal and family history
- Symptom tracking journal
- Thorough physical exam

Some of the laboratory tests that may be done for diagnosis include:
- Antinuclear antibody tests (ANA)
- Autoantibody tests (specific to the condition)
- Complete blood count (CBC)
- C-reactive protein (CRP)
- Erythrocyte sedimentation rate (ESR)
- Comprehensive metabolic panel (CMP)
- Organ function tests and/or imaging (depending on symptoms)


False negative tests may occur.

Because each autoimmune disease is unique, they may be difficult to diagnose. Consult with a specialist or refer and encourage your patient to see one with a focus on their main clinical concerns (see list of referral specialist below).

Treatment Plan

The goals of treatment are to:

- Reduce and relieve symptoms - this can be accomplished with mild medications or supplements for pain. More severe symptoms may need prescription drugs to help relieve symptoms such as pain, swelling, depression, anxiety, sleep problems, fatigue, or rashes. Surgery may be indicated in some instances.
- Control the autoimmune process with antiinflammatories or supplementation of hormones (e.g., insulin and thyroid hormone).
- Maintain the body’s ability to fight disease – by suppressing immune activity to preserve organ function (e.g., chemotherapy, steroids, anti-TNF medications).

Treatments used are dependent on the specific disease and your symptoms and new treatments for autoimmune diseases are currently being researched.

In addition, symptoms may be improved by:

- Eating healthy, well-balanced meals.
- Getting regular moderate physical activity. A gradual and gentle exercise program often works well for people with long-lasting muscle and joint pain.
- Get enough rest. Most people need at least 7 to 9 hours of sleep each day to feel well rested.
• Stress reduction. Stress and anxiety can trigger symptoms to flare up with some autoimmune diseases. Joining a support group or talking with a counselor might also help a patient manage stress and cope with symptoms and disease.

**Referral Criteria**

Specialists may be helpful and necessary in managing some symptoms of autoimmune disease.

• Nephrologist (SLE)
• Rheumatologist (any inflammatory arthropathy or myopathy, scleroderma and SLE)
• Endocrinologist (diabetes and thyroid disease)
• Neurologist (inflammatory myopathy, multiple sclerosis and myasthenia gravis)
• Hematologist (anemia)
• Gastroenterologist. A doctor who treats problems with the digestive system, such as inflammatory bowel disease.
• Dermatologist (psoriasis and SLE)
• Physical therapist (stiffness, weakness, and decreased range of motion)
• Occupational therapist (improve activities of daily living (ADLs)
• Speech therapist (multiple sclerosis)
• Audiologist (inner ear damage from various autoimmune diseases)
• Vocational therapist (job training for people who cannot do their current jobs because of their illness or other health problems)
• Counselor or mental health therapist (coping skills for feelings of anger, fear, denial, and frustration)

**Resources for Clinicians and Patients**


Condition specific association(s)

**The Evidence**


**Clinical Pathway Feedback**

CHP desires to keep our clinical pathways customarily updated. If you wish to provide additional input, please use the e-mail address listed below and identify which clinical pathway you are referencing. Thank you for taking the time to give us your comments.

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