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### A SNAPSHOT GUIDE TO UNDERSTANDING AUTOIMMUNE DISEASE

Having an autoimmune disease is like riding a rollercoaster: One month you're doing fine, the next you're not feeling well at all. And because symptoms come and go and can be vague, getting a diagnosis can take years.

This troubling situation makes it that much more important to take charge of your health. In this guide, we'll explain autoimmunity, describe the key factors that play a role and give a snapshot of six of the most common autoimmune diseases. Plus, we provide a worksheet to help you organize information to discuss with doctors when you're seeking a diagnosis.

Being the biggest champion for your own health starts now.



# WHENYOUR IMMUNE SYSTEM BECOMES OVERACTIVE

"Autoimmune disease occurs when the immune system becomes active in a way that damages normal body structures." — Ana-Maria Orbai, M.D., M.H.S., director of the Psoriatic Arthritis Program at Johns Hopkins



The immune system is a highly sophisticated organization of cells, tissues and organs that work together to protect the body from disease. In many ways, it's like your body's military. It looks for and destroys germs and unhealthy cells, such as cancer. In order to do this, a healthy immune system must tell the difference between beneficial microorganisms, like friendly gut bacteria that help with food digestion, and bacteria and viruses that cause harm.

Your body's defense system has three primary functions:

- Keep watch for foreign substances and unhealthy cells.
- Sound the alert when a threat is found.
- Attack and destroy detected threats.

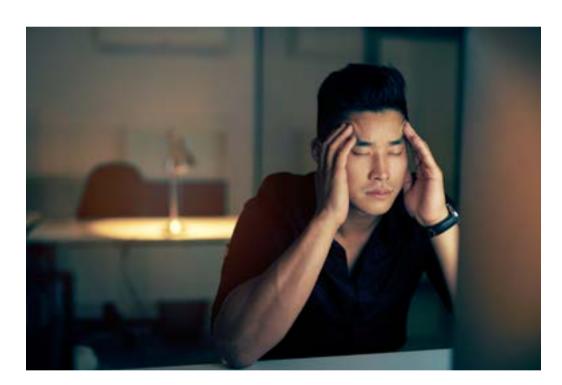


When it's functioning properly, the immune system works to keep you at optimal health. But sometimes something goes wrong and the immune system becomes confused and overzealous. The result is autoimmune disease.

#### A Confused Immune System

"What happens in autoimmune disease is that a real issue such as an infection or injury activates an immune response," explains Ana-Maria Orbai, M.D., M.H.S., director of the Psoriatic Arthritis Program at Johns Hopkins. "But instead of stopping once the threat is contained, the immune system continues functioning as though there's a danger and attacks normal tissue. The reason it goes beyond the scope of its initial job is something we still don't know."

The immune system functions in every part of the body. Thus, autoimmune diseases are wide-ranging and can impact the skin, muscles, joints, vital organs and nervous system.



A key weapon in the immune system's arsenal is inflammation, which signals that an area needs defending or healing. If you're sick, injured or dealing with an autoimmune disease, you can have symptoms of inflammation including:

- Fatigue
- Swelling
- Fever
- Redness
- Muscle aches
- Digestive problems

#### Diagnosis Can Be Challenging

The diagnosis journey is often long and frustrating for those with autoimmune disease. Many of these conditions start with vague and general symptoms that not only overlap with symptoms of other diseases, but that may also come and go.

For most autoimmune diseases, there's not a single test that provides a diagnosis. Instead, doctors have to rule out other illnesses, take blood and tissue samples, and monitor symptoms before they can make a definitive diagnosis.

Consider taking the following steps to help during the diagnostic process:

- Find out if autoimmune disease runs in your family.
- Keep a running list of symptoms to bring to your doctors' appointments.
- Seek additional medical opinions.



#### **Autoimmune Risk Factors**

The exact cause of autoimmune disorders is unknown. However, your genes (inherited family traits) and your lifestyle choices can impact your risk level.

#### The role of genetics

Anyone can develop an autoimmune disease. But if these conditions run in your family, you're more likely to develop one. That's because of genetic material that's passed down from one generation to another. But unlike other inherited diseases that have a direct connection to one or a few specific genes, the link between autoimmune disease and genes isn't so clear-cut.

"Typically, risk level increases from having a multitude of certain genes," explains Orbai. "It's not like breast cancer where you can test for a handful of specific genes to see if you're at higher risk. We're talking about dozens of genes, each one slightly raising the risk of developing an autoimmune disease."

These genetic-based factors increase your risk:

- Family history of autoimmune disease: Indicates it runs in your family.
- Personal history of autoimmune disease: If you have one autoimmune disease, you're at higher risk of developing others.
- **Gender:** 75% of those with autoimmune disease are women, many of whom develop a condition during childbearing years.



#### Lifestyle plays a part

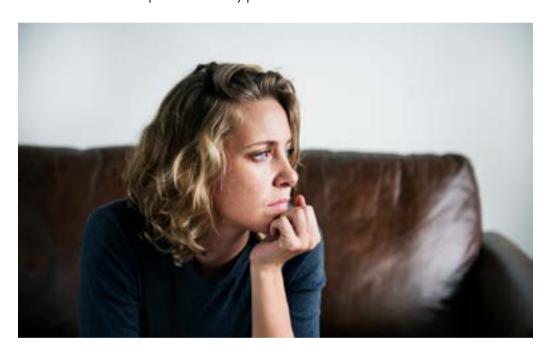
There are steps you can take to decrease your chances of developing an autoimmune disease, even if they run in your family. Certain lifestyle factors contribute to the onset and worsening of autoimmune diseases:

- Obesity: Fat cells secrete substances that trigger an immune response and amplify inflammation. Maintaining a healthy weight decreases inflammation levels.
- Smoking: Studies have linked smoking to the development of multiple autoimmune diseases. Quitting smoking significantly reduces your risk.
- **Stress:** Experiencing high levels of stress is associated with the onset of autoimmune disease. Making lifestyle changes and managing stress promotes good health.

## COMMON AUTOIMMUNE DISEASES

When the immune system turns on itself, it can attack any area of the body. Symptoms of autoimmune disease can be worse for periods of time (called flares) and differ based on what part of the body is involved.

Some autoimmune diseases are extremely rare while others are more common. In this section, you'll find information on six of the most prevalent types of autoimmune disease.



#### **Nearly 100 Autoimmune Diseases**

There are more than 80 different types of autoimmune diseases. Some of the more common ones include:

- Inflammatory bowel disease (IBD)
- Guillain-Barré syndrome
- Celiac disease
- Type 1 diabetes
- Crohn's disease

Learn more about several of the most common <u>autoimmune</u> diseases.

#### **Targeted Treatments**

There are no lasting treatments for autoimmune disease yet, but targeted treatments can reduce and prevent symptoms. "It's expected with autoimmune disease that there will be flares during which you'll experience a worsening of symptoms," says Orbai. "The role of medication is to reduce the intensity of flares and speed up the return of a normal, controlled immune response."

Long used to treat autoimmune disease, immunosuppressive drugs subdue the entire immune system, making it less responsive in general. That can make those with autoimmune disease prone to infections and other disease. But a new generation of medications called biologic drugs have a more targeted approach to halting symptoms. Biologic drugs take aim at singular points in the immune response process to prevent symptoms from occurring.

"The immune system is very delicate and not fully charted," says Orbai. "Interfering with it has risks and benefits. The more targeted the medication, the less disruptive it is to the body's overall defense."



#### Systemic Lupus Erythematosus (Lupus)

#### What is it?

As the name implies, systemic lupus erythematosus is a bodywide autoimmune disease. It usually affects the skin, joints and kidneys but can also impact the brain, heart, lungs and blood cells.



#### What are the most common symptoms?

- Extreme fatigue
- Headaches
- Low-grade fever
- Joint stiffness
- Chest pain with deep breathing
- Swelling in the hands, feet, legs and around the eyes
- Rashes on sun-exposed skin, including a butterfly-shaped rash (called a malar rash) over the nose and cheeks

#### How is it treated?

There are multiple types of medications used to reduce lupus symptoms, including:

- **Biologic drugs:** These newer medications work by blocking part of the immune response that leads to excess inflammation.
- **Steroids and non-steroidal drugs:** These drugs are used to decrease existing inflammation.
- Anti-malarial drugs: Originally designed to treat malaria, these medications also control lupus flares and lessen damage inflicted by an overactive immune system.
- Immunosuppressive drugs: These drugs are used for more severe cases of lupus. They lower the immune system's overall defenses. But by doing so, they also increase the risk of infection and cancer.



Learn more about <u>lupus</u> and how it's diagnosed and treated at the <u>Johns Hopkins Lupus Center</u>.

#### **Psoriasis**

Psoriasis is an autoimmune disease that triggers abnormally rapid production of skin cells, which build up on the skin surface and create irritation. There are multiple types of psoriasis, but the most common is plaque psoriasis, which causes thick, scaly patches of skin. Psoriasis can also affect nails or joints. Joint-related psoriasis is called psoriatic arthritis.

#### What are the most common symptoms?

- Thickened patches of skin with silvery scales (called plaques)
- Itching or burning sensation in affected areas
- Plaques on the elbows, knees, lower back or scalp
- Thickened or pitted nails
- In children, small red spots or raised skin
- Small pus-filled blisters
- Stiff, painful joints

#### How is it treated?

There are a variety of psoriasis treatments that target skin cell overproduction and inflammation, such as:

- Over-the-counter options: For mild psoriasis, treatments containing salicylic acid or coal tar can reduce scaling. Thick moisturizers can ease dryness.
- **Topical prescription medications:** Creams or liquids with steroids or derivatives of vitamin A or vitamin D are applied to the skin to normalize cell growth and reduce inflammation.

- **Ultraviolet (UV) light therapy:** Exposure to UV rays, sometimes coupled with medication that makes skin more sensitive to light, is used to treat mild to severe psoriasis.
- **Biologic drugs:** These drugs block part of the immune response to reduce psoriasis symptoms.
- Immunosuppressive drugs: For severe psoriasis, these drugs lower the immune system's overall defenses. But by doing so, they also increase the risk of infection and cancer.



Learn more about <u>psoriasis</u> and how psoriatic arthritis it is treated at the <u>Johns Hopkins Arthritis Center</u>.

#### Multiple Sclerosis (MS)

In multiple sclerosis, the immune system turns on the brain and spinal cord. It attacks the protective lining around nerves, leaving them exposed and disrupting communication between the brain and the body.

#### What are the most common symptoms?

- Fatigue
- Dizziness
- Slurred speech
- Incontinence or constipation
- Tingling, numbness, weakness or pain in parts of the body
- Loss of coordination or control over parts of the body

#### How is it treated?

• **Steroids:** During an MS flare-up, steroids are used for a brief time to make the immune system's attack on the nervous system shorter and less severe.

- **Biologic drugs:** These drugs are injected or given intravenously as a preventive therapy to block part of the immune response and lessen MS flares.
- **Rehabilitation:** Physical and occupational therapy help with regaining or maintaining optimal functioning.



Learn more about <u>multiple sclerosis</u> and how it is treated at the Johns Hopkins <u>Multiple Sclerosis Center</u>.

#### Rheumatoid Arthritis (RA)

This autoimmune disease causes the lining of joints to become inflamed. Inflammation typically happens on both sides of the body at the same time (for example: both wrists or both hands). Left unchecked, the joint damage from rheumatoid arthritis leads to deformity as joints loosen and shift. In some cases, RA may also affect the heart, lungs or eyes.

#### What are the most common symptoms?

- Pain or aching in more than one joint
- Stiffness or swelling in multiple joints (worse in the morning)
- General feeling of weakness or fatigue
- Low-grade fever
- Loss of appetite

#### How is it treated?

- **Steroids and non-steroidal drugs:** These drugs are used to decrease inflammation and reduce pain. Steroids are only used for a brief period of time.
- Immunosuppressive drugs: Known as diseasemodifying anti-rheumatic drugs (DMARDs), these drugs

lower the immune system's overall defenses to prevent damaging inflammation. However, they also increase the risk of infection and cancer.

• **Biologic drugs:** As a preventive therapy, these drugs are injected or given intravenously to block a specific part of the immune response and can also increase the risk of infections.



Learn more about <u>rheumatoid arthritis</u> and how it is treated at the <u>Johns Hopkins Arthritis Center</u>.

#### Myasthenia Gravis

Nerves send messages to voluntary muscles (the ones you can control) to get them to act. In myasthenia gravis, the immune system blocks the communication between nerves and muscles. As a result, muscles don't move. The symptoms of the disease vary greatly, sometimes improving or getting worse within a matter of hours.

#### What are the most common symptoms?

- Muscle weakness that worsens after activity and improves with rest
- Drooping of one or both eyelids and/or double vision
- Change in facial expression
- Difficulties speaking or voice changes
- Weakness in the limbs, hands, fingers or neck
- Trouble chewing or swallowing, which can lead to choking or coughing during meals

#### How is it treated?

- **Thymus removal:** Surgery to remove the thymus gland, which plays a role in myasthenia gravis, often reduces symptoms. Sometimes no additional treatment is needed.
- Immunosuppressive drugs: These drugs suppress immune system activity to reduce attacks on nerve signals. But by doing so, they also lower the body's overall defense and may cause side effects such as nausea, diarrhea and vomiting.
- Anti-cholinesterase drugs: These medications improve nerve-to-muscle communication and increase muscle strength.
- Plasmapheresis and intravenous immunoglobulin: In myasthenia gravis, the immune system makes antibodies that attack the communication between nerves and muscles. Two treatments remove these antibodies from the blood. Plasmapheresis uses a machine to filter harmful antibodies out of the blood while intravenous immunoglobulin uses an injection of healthy donor antibodies that take harmful antibodies out of circulation.



Learn more about myasthenia gravis and how it is treated.

## Autoimmune Thyroid Disease: Graves' Disease & Hashimoto's Disease

Autoimmune thyroid disease (AITD) occurs when the immune system attacks the thyroid, a gland in the neck that secretes hormones to control the functioning of multiple parts of the body. Due to damage caused by the immune system, the thyroid loses its ability to properly function. This can result in two different autoimmune diseases, both of which affect the way

energy is used in the body by various organs, including the heart.

- **Hashimoto's disease:** Damage to the thyroid makes it unable to produce enough hormones (referred to as hypothyroidism).
- **Graves' disease:** Damage to the thyroid may cause it to make more hormones than the body needs (referred to as hyperthyroidism).

#### What are the most common symptoms?

#### Hashimoto's disease:

- Fatigue
- Weight gain
- Constipation
- Dry skin and hair and hair loss
- Increased sensitivity to cold
- Achy joints or muscles
- Heavy or irregular menstrual periods

#### Graves' disease:

- Racing and irregular heartbeat
- Frequent and loose stools
- Increased sensitivity to heat
- Unexpected weight loss
- Trembling hands
- Trouble sleeping
- Muscle weakness

#### How is it treated?

• Thyroid hormone medications: The treatment for Hashimoto's disease is a daily medication that replaces the hormones the thyroid isn't producing.

- **Radioiodine therapy:** The most common treatment for Graves' disease is usually one dose of radioactive iodine-131, which slowly shuts down the thyroid's ability to produce hormones.
- **Beta blockers:** These drugs are used to treat some of the most concerning symptoms of Graves' disease, including rapid heartbeat and trembling.
- Anti-thyroid drugs: For people with Graves' disease, these medications reduce how much hormone the thyroid can make. However, the results only work for a limited time and can cause side effects such as increased risk of infection.

LEARN MORE Learn more about <u>thyroid disorders</u> in women and how they are treated.



#### Talking Regularly with Your Doctor Is Key

Staying in regular contact with your doctor is a crucial part of managing autoimmune disease. "Figuring out the best treatment for you at various stages is a fine-tuning process," explains Orbai. "Doctors and patients have to work together to reach and maintain a balance of controlling symptoms and side effects."

Autoimmune diseases can change over time — triggering stronger immune responses or going into remission — and you can also develop a tolerance to medications you're taking. If your symptoms have gotten worse, it might be a sign that your condition is not well controlled. You may need more medication or a different one. Alternatively, if your symptoms disappear, your doctor may want to reduce your medication.

The bottom line: Keeping in touch with your doctor is essential to making sure you're getting the best possible treatment.



## PREPARING FOR YOUR APPOINTMENT

#### MEDICALTRACKER

Use this sheet to keep track of pertinent information to bring with you to doctors' appointments.

Symptoms:
Personal Medical History:
Note previous tests, diagnoses and treatments.
Family History:
Note medical history of close blood relatives.

Questions:	
Appointment Log:	
Date:	
Doctor:	
Location:	
Discussion:	
Date:	
Doctor:	
Location:	
Discussion:	
Date:	
Doctor:	
Location:	
Discussion:	

## RESOURCES

<u>Autoimmune Diseases</u> – Information from Johns Hopkins Medicine

Johns Hopkins Arthritis Center

Johns Hopkins Lupus Center

Johns Hopkins Multiple Sclerosis Center