Autoimmune Diseases

Autoimmune diseases result from a dysfunction of the immune system. The immune system protects you from disease and infection. Sometimes, though, the immune system can produce autoantibodies that attack healthy cells, tissues, and organs. This can lead to autoimmune disease.

Autoimmune diseases can affect any part of the body. More than 80 autoimmune diseases have been identified. Some are relatively well known, such as type 1 diabetes, multiple sclerosis, lupus, and rheumatoid arthritis, while others are rare and difficult to diagnose.

Autoimmune diseases are becoming increasingly prevalent, for reasons unknown. Some autoimmune diseases are life-threatening, and most are debilitating and require a lifetime of treatment. There are treatments available to reduce the symptoms and effects from many autoimmune diseases, but cures have yet to be discovered. Since most autoimmune diseases are rare, patients can often spend years seeking a proper diagnosis.

Individually Rare, Collectively Common

Collectively, autoimmune diseases are among the most prevalent diseases in the U.S., affecting more than 23.5 million Americans. They are more common among women, and while some are more prevalent among white people, others are more common among African-Americans and Hispanics. Autoimmune diseases are becoming increasingly prevalent, for reasons unknown.

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The causes of autoimmune diseases remain largely unknown. There is growing consensus that autoimmune diseases likely result from interactions between genetic and environmental factors. The National Institute of Environmental Health Sciences (NIEHS) is supporting research to understand how these factors work together to compromise the body’s ability to defend itself, and develop into autoimmune diseases. NIEHS hopes to find clues that will lead to treatments and cures, or ways to prevent the development of these diseases.

More than 80 autoimmune diseases have been identified. Some of the more common ones include:

- Diabetes (Type 1)
- Lupus
- Multiple sclerosis
- Rheumatoid arthritis
- Celiac disease

NIEHS and NTP Research Efforts

Unraveling the connections between genetic predisposition and environmental triggers is a major focus for NIEHS and the National Toxicology Program (NTP), an interagency testing program headquartered at NIEHS. Progress is being made through multiple research efforts, some of which are noted below.

- A 2012 study by NIEHS researchers found that over 32 million people in the U.S. have autoantibodies. Earlier studies have shown that autoantibodies can develop many years before the clinical appearance of autoimmune diseases. The study, which looked at the most common autoantibodies, antinuclear antibodies, found that they are most prevalent among women. This research suggests that the hormones estrogen and progesterone might be affecting the immune system.

- A study of residents in Libby, Mont., who have experienced significant exposure to asbestos minerals due to mining in the area, suggested a link between asbestos exposure and lesions in the lungs. Sixty-one percent of Libby residents tested had autoantibodies and were more likely to have two types of lung abnormalities.

- An NIEHS study found associations between ultraviolet radiation from sunlight and the development of an autoimmune muscle disease, myositis, particularly in women.

- Low birth weight and low socioeconomic factors in childhood were associated with the later development of rheumatoid arthritis as an adult.
• Recognizing that individuals are never exposed to one chemical at a time, NIEHS grantees studied what happens when mice are exposed to two suspected triggers for autoimmune diseases. Previous studies had shown that exposure to trichloroethylene, a solvent and degreasing compound, induced autoimmune hepatitis in autoimmune-prone mice. This study found that when the mice were also exposed to mercuric chloride, a compound used as a disinfectant and also in photography, disease development accelerated and a unique, liver-specific autoantibody response occurred.6

• NIEHS grantees studying blood samples of Brazilian mothers exposed to methylmercury, an environmental contaminant passed on to humans by eating contaminated fish, found elevated levels of autoantibodies in the blood of both mothers and their fetuses.7

• NIEHS and NTP researchers demonstrated that a certain enzyme creates mutations in DNA and is a major player in the development of autoantibodies. The discovery of the role of this enzyme establishes it as a potential target for therapy in autoimmune disorders, such as lupus.8

The findings include:
• Exposure to solvents, which are used in thousands of products, including paint thinners, cleaning supplies, and nail polish, contributes to the development of systemic sclerosis.

• Smoking contributes to the development of two types of rheumatoid arthritis.

• Exposure to fine particles of crystalline silica, a basic component of quartz, granite, and many other minerals, contributes to the development of several autoimmune diseases. Workers exposed to these minerals are particularly at risk.

• Eating gluten, present in wheat and some other grains, contributes to the development of celiac disease, a disorder that affects the small intestine and commonly causes chronic diarrhea and fatigue.

• Taking certain supplements containing L-tryptophan, an essential amino acid used as a dietary supplement, contributes to development of eosinophilia myalgia syndrome, an incurable and sometimes fatal condition involving severe muscle pain.

**Unraveling Environmental Triggers**

NIEHS brought together an interdisciplinary group of experts to evaluate the state of the science regarding the role of the environment and the development of autoimmune diseases.

The experts identified future research directions, identified promising mechanistic theories and animal models, and identified some specific environmental agents that may be involved in the development of autoimmune diseases.9,10,11

![A complex interplay of genes and environment lead to autoimmune diseases.](image-url)

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