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Autoimmune Diseases and Laboratory Diagnosis - Dr. K. Leena MD

An autoimmune disease develops when the immune system, which defends the body against disease, decides the healthy cells are “foreign”. As a result, the immune system attacks the healthy body cells, causing tissue destruction. This immune attack – which may be generalised and take place in a number of tissues, or may focus on a single organ – is executed by substances called autoantibodies, which target specific components of cells such as nucleus or cell receptors.

There are as many as 80 types of autoimmune diseases. Many of them have similar symptoms, which makes them very difficult to diagnose. It is also possible to have more than one at the same time. They usually fluctuate between periods of remission (little or no symptoms) and flare-ups (worsening symptoms). There are no cures for autoimmune diseases, so treatment focusses on relieving the symptoms. Autoimmune diseases often run in families and 75 percent of those affected are women.

SOME COMMON AUTOIMMUNE DISEASES

The following are some of the more common autoimmune diseases:

- **Rheumatoid arthritis** – inflammation of joints and surrounding tissues.
- **Systemic lupus erythematosus (SLE)** – affects skin, joints, kidneys, brain, and other organs.
- **Multiple sclerosis** – affects the brain and spinal cord.
- **Celiac sprue disease** – a reaction to gluten (found in wheat, rye, barley) that causes the damage to the lining of the small intestines.
- **Pernicious anaemia** – decrease in red blood cells caused by inability to absorb vitamin B12.
- **Vitiligo** – white patches on the skin caused by loss of pigment.
- **Scleroderma** – a connective tissue disease that causes changes in skin, blood vessels, muscles, and internal organs.
- **Psoriasis** – a skin condition that causes redness and irritation as well as thick, flaky, silver-white patches.
- **Hashimoto’s disease** – inflammation of the thyroid gland.
- **Addison’s disease** – adrenal hormone deficiency.
- **Grave’s disease** – overactive thyroid gland.
- **Reactive arthritis** – inflammation of joints, urethra, and eyes; may cause sores on the skin and mucus membranes.
- **Type 1 diabetes** – destruction of insulin producing cells in the pancreas.

WHAT CAUSES THE IMMUNE SYSTEM TO ATTACK HEALTHY BODY CELLS?

The cause of autoimmune disease is unknown. There are many theories about what triggers autoimmune diseases, including

- Bacteria or virus.
- Drugs.
- Chemical irritants.
- Environmental irritants.

WHAT ARE THE SYMPTOMS OF AN AUTOIMMUNE DISEASE?

Because there are many different types of autoimmune disease, the symptoms may vary. However, most of them cause fatigue, fever, and general malaise (feeling ill). Symptoms worsen during flare-ups and lessen during remission. Autoimmune diseases attack many parts of the body. The most common organs and tissue affected are:

- Joints.
- Muscles.

- Skin.
- Red blood cells.
- Blood vessels.
- Connective tissue.
- Endocrine gland.

HOW ARE AUTOIMMUNE DISEASES DIAGNOSED?

Ordinarily, the immune system produces antibodies (proteins that recognise and destroy specific substances) against harmful invaders in the body, such as viruses, bacteria, parasites and fungi. When one has an autoimmune disease, the body produces antibodies against some of the body's own tissues. Diagnosing an autoimmune disease involves identifying the antibodies produced by the body. The following tests are used to diagnose an autoimmune disease:

- **ANTINUCLEAR ANTIBODY (ANA) TESTS** – A type of autoantibody test that looks for antinuclear antibodies, which attacks the nucleus of the cells.
- **AUTOANTIBODY TESTS** – Any of several tests that look for specific antibodies to one's own tissues.
- **COMPLETE BLOOD COUNT (CBC)** – Measures the numbers of red and white cells in the blood. When the immune system is actively fighting something, these numbers will vary from the norm.
- **C-REACTIVE PROTEIN (CRP)** – Elevated CRP is an indication of inflammation in the body.
- **ERYTHROCYTE SEDIMENTATION RATE (ESR)**
- **RHEUMATOID FACTOR (RF or Rh Factor)** – Like ANA, it can detect an abnormal protein that the immune system makes when attacking the body.
- **ANTI-CYCLIC CITRULLINATED PEPTIDE ANTIBODY TEST (ANTI-CCP)** - The test is usually done along with Rh Factor test to confirm a diagnosis of rheumatoid arthritis.

SPECIAL CONCERNS ABOUT ANTIBODY TESTS

- False-positive and false-negative results are possible.
- Certain medications may alter the results of some of these tests.
- A radioactive scan performed within one week before certain tests may alter the result.
- Antinuclear antibodies may be found in the blood of healthy older people and occasionally in some healthy younger individuals.

ANTIBODY TEST RESULTS

The blood sample is sent to a laboratory and analysed for the presence of autoantibodies. The doctor will review the test results in conjunction with symptoms, medical history, and physical examination for evidence of an autoimmune disorder.

If results are positive and the doctor can make a definitive diagnosis, appropriate therapy may begin.

In many cases, one or more positive results on these blood tests will necessitate additional procedures – for example, a biopsy of the skin, liver or muscle - to confirm a diagnosis.

The absence of autoantibodies suggests one may have a condition other than an autoimmune disorder, and other tests may be scheduled.

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