

Rare Genomics-Genetic Diseases & Autoimmune Diseases

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An autoimmune disease is a condition arising from an abnormal immune response to a functioning body part. There are at least 80 types of autoimmune diseases. Nearly any body part can be involved. Common symptoms include low grade fever and feeling tired. Often symptoms come and go. The cause is generally unknown. Some autoimmune diseases such as lupus run in families, and certain cases may be triggered by infections or other environmental factors. Some common diseases that are generally considered autoimmune include celiac disease, diabetes mellitus type 1, Graves' disease, inflammatory bowel disease, multiple sclerosis, psoriasis, rheumatoid arthritis, and systemic lupus erythematosus. The diagnosis can be difficult to determine. Treatment depends on the type and severity of the condition. Nonsteroidal anti-inflammatory drugs (NSAIDs) and immunosuppressants are often used. Intravenous immunoglobulin may also occasionally be used. While treatment usually improves symptoms, they do not typically cure the disease. About 24 million (7%) people in the United States are affected by an autoimmune disease. Women are more commonly affected than men. Often they start during adulthood. The first autoimmune diseases were described in the early 1900s.

Signs and symptoms:

Autoimmune diseases present similar symptoms across the more than eighty different types. The appearance and severity of these signs and symptoms depends on the location and type of autoimmune response that occurs. An individual may also have more than one autoimmune disease simultaneously, and display symptoms of multiple diseases. Signs and symptoms presented, and the disease itself, can be influenced by various other factors such as age, hormones, and environmental factors. In general, the common symptoms are: Fatigue, Low grade fever, General feeling of unwell, Muscle aches and joint pain and Rash on different areas of the skin. The appearance of these signs and symptoms can fluctuate, and when they reappear, it is known as a flare-up. Such signs and symptoms may aid in diagnosis by supporting the results from biologic markers of autoimmune diseases. There are several areas that are commonly impacted by autoimmune diseases. These areas

include: blood vessels, underlying connective tissues, joints and muscles, red blood cells, skin, and endocrine glands, like thyroid or pancreas glands. These diseases tend to have characteristic pathological effects that characterize them as an autoimmune disease. Such features include damage to or destruction of tissues where there is an abnormal immune response, altered organ growth, and altered organ function depending on the location of the disease. Some diseases are organ specific and are restricted to affecting certain tissues, while others are systemic diseases that impact many tissues throughout the body. Signs and symptoms may vary depending on which of these categories an individual's disease falls under. Coeliac Disease: Coeliac disease presents the strongest associations to gastrointestinal and lymphoproliferative cancers. In coeliac disease, the autoimmune reaction is caused by the body's loss of immune tolerance to ingested gluten, found primarily in wheat, barley, and rye. This explains the increased risk of gastrointestinal cancers, as the gastrointestinal tract includes the esophagus, stomach, small intestine, large intestine, rectum, and anus, all areas that the ingested gluten would traverse in digestion. The incidence of gastrointestinal cancer can be partially reduced or eliminated if a patient removes gluten from their diet. Additionally, celiac disease is correlated with lymphoproliferative cancers. Inflammatory Bowel Disease: Inflammatory bowel disease is associated with cancers of the gastrointestinal tract and some lymphoproliferative cancers. Inflammatory bowel disease (IBD) can be further categorized as Crohn's disease or ulcerative colitis. In both cases, individuals with IBD lose immune tolerance for normal bacteria present in the gut microbiome. In this case, the immune system attacks the bacteria and induces chronic inflammation, which has been linked to increased cancer risk. Multiple Sclerosis: Multiple sclerosis is associated with decreased risk of cancer overall but an increased risk of central nervous system cancer, primarily in the brain. Multiple sclerosis is a neurodegenerative disease in which T-cells – a specific type of immune cells – attack the important myelin sheath in brain neurons. This reduces the nervous system function, creating inflammation and subsequent cancer of the brain.