CONNECTIVE TISSUE DISORDERS

Collagen and Rheumatological Disorders and Their Causes

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The autoimmune connective tissue disorders include various organ-specific and systemic conditions such as lupus and Sjogren's that affect structural tissue.

The Connective Tissue Disease Family

Connective tissue disorders include a family of more than 200 different disorders that affect connective tissues. Connective tissue disorders are caused primarily by gene mutations affecting the production of tissue and by a number of different specific and overlapping autoimmune diseases.

Systemic vs Organ-specific diseases

In autoimmune connective tissue disorders, specific organs (organ-specific diseases) or multiple organs (systemic diseases) may be affected. Up until the late 70's, most systemic or rheumatological autoimmune diseases were referred to as connective tissue diseases or collagen diseases. Today, connective tissue diseases are classified as either 1) autoimmune connective tissue disorders such as lupus disorders, rheumatoid arthritis, and dermatomyositis or 2) heritable connective tissue disorders (HCTDs) such as Ehlers-Danlos syndrome, epidermolysis bullosa, and Marfan syndrome caused by gene mutations. In the HCTDs, alterations in affected genes may change the structure and development of connective tissue in specific organs.

Connective Tissue

Connective tissue is a glue-like material or matrix that connects the cells that make up the body’s tissues. Connective tissue gives the body’s tissues and organs strength, form, and flexibility. Connective tissue also provides nutrients to tissue and aids in the special functions of certain tissues. For instance, connective tissue in joints gives them the ability to move. Connective tissue is composed of dozens of proteins and compounds containing various combinations of protein and glucose. Connective tissues' component proteins include collagens, elastins, proteoglycans, and glycoproteins. Various combinations of these proteins are found in different tissues. For instance the connective tissue in ligaments is primarily composed of elastin. Connective tissue is found in many different organs, including the skin, bones, joints, heart, blood vessels, lungs, eyes and ears.

Autoimmune Connective Tissue Disorders

The autoimmune connective tissue disorders include a number of specific conditions and overlap syndromes including:
* Rheumatoid arthritis (RA)
* Systemic lupus erythematosus (SLE or lupus)
* Polymyositis
* Dermatomyositis
* Systemic sclerosis (scleroderma)
* Sjogren’s syndrome and Sjogren's subtypes
* Various forms of vasculitis including temporal arteritis, Wegener's granulomatosis, and central nervous system vasculitis
* Mixed connective tissue disease
* Undifferentiated connective tissue disease
* Raynaud’s syndrome and disease
* Lupus overlap syndromes
* Autoimmune overlap syndromes

**Disease Course and Symptoms**

Females are more likely to be affected than males, and although the prime age is 30-50 years, people of all ages may be affected. Most of the autoimmune connective tissue disorders affect multiple tissues, and the blood vessels are the organs most often affected. Many of the specific autoimmune connective disorders tend to overlap or appear in conjunction with other connective tissue disorders. For instance, patients with SLE may have joint problems characteristically seen in rheumatoid arthritis. The autoimmune connective tissue disorders may develop slowly over many years or they may present abruptly and show rapid progression, and they're typically characterized by alternating periods of remission and flares.

Symptoms in autoimmune connective disorders may affect the function of specific organs such as the kidney involvement that often occurs in SLE or the joint pain characteristic of rheumatoid arthritis. Pain, inflammation, tenderness, dryness and irritation of mucous membranes, and rashes and hives are all prominent features.

Specific connective tissue disorders cause specific symptoms, and specific disorders are associated with the presence of specific antinuclear antibody patterns and circulating blood levels of rheumatoid factor. Symptoms, autoantibody test results, inflammatory
blood markers, and imaging tests are used to diagnose specific conditions. However, antinuclear (ANA) test results are sometimes inconclusive and there are known pitfalls to ANA testing. When an absence of classic symptoms and autoantibody patterns make diagnosis difficult, patients are said to have undifferentiated connective tissue disease. Undifferentiated connective tissue disorders may eventually develop into classic connective tissue diseases or they may remain undefined.

**Resources:**


Thomas Medgser, Lupus in Overlap with Other Connective Tissue Diseases, Lupus Foundation of America, www.lupus.org/education/brochures/connective.html

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