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Spotlight on Health

Autoimmune Rheumatic Diseases

Autoimmune rheumatic diseases (ARDs) are a diverse group of conditions that primarily affect the joints, bones, muscle, and connective tissue (see Sidebar).[1] They can be especially challenging to diagnose during early stages, often presenting with nonspecific symptoms and signs that may flare and remit. However, early diagnosis is important to improve outcomes for patients.

This newsletter will review ARDs and the importance of early and accurate diagnosis. It will also discuss the importance of laboratory testing for autoantibodies and how antibody panels can help to rule in or rule out common autoimmune diseases.

Characteristics, Scope, and Causes

The ARDs are characterized by an abnormal immune response to normal cells and tissues.[1,2] They can lead to severe, debilitating pain and progressive disease. Some are associated with increased mortality. The abnormal immune response can be directed at a specific part of the body, such as the joints in rheumatoid arthritis (RA).[2] It can also be more general and systemic and affect multiple organs and tissues, such as in systemic lupus erythematosus (SLE).[2] The majority of rheumatic diseases are due to an autoimmune response,[1] whereas others (eg, gout and osteoarthritis) have different pathophysiology.

The overall prevalence of autoimmunity in the general population is approximately 3% to 5%.[2] Women get autoimmune diseases at a rate of more than twice that of men (6.4% of women compared to 2.7% of men).[3] Different autoimmune diseases can occur at different times in a person's life. However, women are most commonly diagnosed with an autoimmune disease when they are between 14 and 44 years old.[3]

Some ARDs, such as SLE, have a genetic component and others can be triggered by infections or environmental factors.[2] However, the cause of an ARD in a specific individual usually cannot be determined.

Diagnosis and Management

Some ARDs have characteristic findings that can aid in diagnosis. For example, the primary symptoms of Sjögren syndrome are dry mouth and dry eyes.[4] Other ARDs have characteristic radiographic findings.[5-7]

However, pathognomonic signs are the exception rather than the rule. The early symptoms of autoimmune diseases can be similar to one another, may flare and remit, and overlap with those of other conditions, including other ARDs. They may include[2,8]

- Muscle aches
- Fatigue
- Swelling and redness over the joints
- Mild fever
- Trouble concentrating
- Rashes
- Hair loss
- Numbness and tingling in the hands and feet

Examples of More Common Autoimmune Rheumatic Diseases

- Rheumatoid arthritis (RA)
- Systemic lupus erythematosus (SLE)
- Sjögren syndrome
- Polymyositis
- Systemic sclerosis
- Mixed connective tissue disease (MCTD)
- CREST (calcinosis, Raynaud phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia) syndrome
- Neuropsychiatric SLE

Consequently, diagnosis of ARDs may be delayed, and irreversible joint or organ damage may ensue. Early diagnosis and treatment provides an opportunity to prevent or mitigate this damage, control symptoms, and improve chances for remission.[2,8]

Autoantibody testing is an important aid in the diagnosis of ARDs.[2,9] Almost all ARDs have specific autoantibody profiles. As such, testing for specific autoantibodies can help rule in or rule out specific ARDs. The use of antibody panels (testing for a number of autoantibodies at the same time) may result in earlier diagnosis, and thus potentially better outcomes for patients.

No single therapy exists for ARDs. Treatments include nonsteroidal anti-inflammatory drugs, immunosuppressants, and cytokine antagonists.[2] In general, autoimmune conditions are not cured; treatments are aimed at minimizing symptoms, inhibiting inflammation, slowing progression, and/or inducing remission.[2]

How the Laboratory Can Help

Quest Diagnostics offers tests for the many different antibodies that are associated with ARDs and other autoimmune diseases. Panels are also available to assist in rapidly ruling in or ruling out certain conditions. For example, the ANA IFA with Reflex to Titer and Reflex to Multiplex 11 Ab Cascade plus IdentRA® panel (test code 94954) assists in the diagnosis of the more common ARDs (see Sidebar on previous page). Quest also offers individual tests for the panel components, as well as more limited but focused
panels for diagnosis of ARDs.

Additional information about testing for ARDs is available at

- QuestDiagnostics.com/test/test-guides/CF_Autoimmune_Rheumatic_Diseases/autoimmune-rheumatic-and-related-diseases?p=r
- QuestDiagnostics.com/test/test-guides/TS_ANA_RflTiterPattern_Rfl11AbCascade_IdentRA/ana-screen-ifa-reflex-titerpattern-reflex-mplx-11-ab-cascade-with-identra?p=td
- QuestDiagnostics.com/test/test-guides/TG_Autoimmune_AbTesting/autoimmune-diseases-use-of-antinuclear-and-specific-antibodies-for-diagnosis?p=r

Model used for illustrative purpose.

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