

Spotlight on Health

Autoimmune Rheumatic Diseases

Autoimmune rheumatic diseases (ARDs) can be challenging to diagnose. They often present with non-specific symptoms and signs, especially during early stages. Early diagnosis, however, is important to improve outcomes.

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.

Autoimmune Rheumatic Diseases

ARDs are a group of conditions characterized by an abnormal immune response to a normal part of the body.¹ They can lead to severe, debilitating pain and progressive disease. Some are associated with increased mortality. The abnormal response can be directed at a specific part of the body, as in rheumatoid arthritis (RA). It can also be more general and affect multiple organs and tissues, such as in systemic lupus erythematosus (SLE).¹

Some of the more common ARDs are RA, Sjögren syndrome, mixed connective tissue disease (MCTD), and psoriasis.² The overall prevalence of autoimmunity in the general population is approximately 3% to 5%.¹ Women get autoimmune diseases at a rate of about 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. Women are most commonly diagnosed with an autoimmune disease when they are between 14 and 44 years old.³

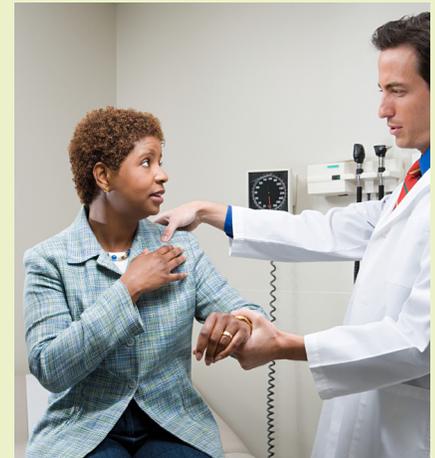
The cause of an ARD in a specific individual usually cannot be determined. However, some conditions such as SLE have a genetic component, and others can be triggered by infections or environmental factors.¹

Diagnosis and Management

Some ARDs, such as psoriasis, have characteristic lesions that can aid in diagnosis. Others do not have specific symptoms and can only be confirmed with common laboratory testing. Still others have characteristic radiographic findings. For example, radiographic characteristics of RA are soft tissue swelling, periarticular osteopenia, joint space narrowing, and marginal erosions.⁴ Characteristic features are also identifiable on magnetic resonance imaging.⁴

However, pathognomonic signs are the exception rather than the rule. For most ARDs, symptoms can wax, wane, and overlap with those of other conditions, including other ARDs (sidebar).³ This can delay diagnosis.⁵ Early diagnosis, however, is important: many of these conditions can cause irreversible organ damage, which could be prevented or mitigated with timely treatment.⁶ Chances for remission or symptom control are also improved with prompt diagnosis and treatment.⁶

Autoantibody testing is the mainstay to aid in the diagnosis of ARDs.¹ Almost all ARDs have specific autoantibody profiles. As such, testing for specific



Symptoms of Autoimmune Diseases

The early symptoms of autoimmune diseases can be similar to one another. They may include³:

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

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, a better outcome.

There is no single therapy for ARDs. Treatments include nonsteroidal anti-inflammatory drugs, immunosuppressants, targeted therapies, and cytokine antagonists.⁵ In general, autoimmune conditions are not cured; treatments are aimed at minimizing symptoms and/or inducing remission.⁵

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 RA, CREST (calcinosis, Raynaud phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia) syndrome, SLE and neuropsychiatric SLE, Sjögren syndrome, systemic sclerosis, polymyositis, and MCTD. Quest Diagnostics test code 16814 (ANA Screen, IFA, with Reflex Titer/Pattern, and Reflex to Multiplex 11 Antibody Cascade) can be used to test for all of the same conditions, with the exception of RA.

References

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