



AUTOIMMUNITY AUTOANTIBODIES AND DISORDERS

Autoimmunity is a complex phenomenon where the immune system, which is normally tasked with protecting the body from foreign pathogens, mistakenly turns on the body itself. Such dysfunction may be due to a variety of factors, particularly **genetic**, **environment and hormonal**.

Autoimmune conditions, which affect millions of people across the world, have significant consequences for public health. Indeed, epidemiological data show that autoimmune diseases affect up to **5 to 8% of the world population**, and their prevalence is ever increasing. Thus, the development of strategies for prevention, early diagnosis and effective treatment are key elements in modern medicine.

Autoimmune disorders

Autoimmune disorders present in different ways, and may affect a wide range of organs and tissues. They are typically classified into two categories. **Non-organ specific disorders**, which are systemic, with multifocal expression and the involvement of several organs. **Organ specific disorders**, when the clinical expression involves a single target organ.

(1) Non-organ specific autoimmune disorders

These conditions mainly include connective tissue and some vascular tissue diseases. They are very different from each other in their clinical presentation, yet share some common characteristics.

COMMON CLINICAL CRITERIA Systemic signs • Fever • Asthenia • Myalgia	 CONNECTIVE TISSUE DISEASES Systemic lupus erythematosus Rheumatoid arthritis Sjögren syndrome Scleroderma Polymyositis Dermatomyositis Mixed connective tissue disease 	Test for antinuclear antibodies (ANAs)	
Joint pain • Arthritis • Arthralgia	 VASCULAR TISSUE DISEASES Polyarteritis nodosa (PAN) Granulomatosis with polyangiitis (GPA) Microscopic polyangiitis (MPA) Eosinophilic granulomatosis with polyangiitis (EGPA) 	Test for anti- neutrophil cytoplasmic antibodies (ANCAs	ی چي کي کي

In spite of the classification according to clinical or biological criteria, the clinical diagnosis is often difficult, since only one out of five patients presents typical clinical signs or evocative symptoms. Thus, **an autoantibody test is indispensable** for a differential diagnosis.

2 Organ specific autoimmune disorders

These conditions are many and vary depending on the affected organ. Each condition has specific autoantibody panels. In the event of a clinical suspicion, these panels should be prescribed, with the indication "autoantibody test" followed by the suspected syndrome (e.g.:search for autoantibodies for diabetes, myasthenia, primary biliary cholangitis etc.).



Value of autoantibody tests

Autoantibody testing takes on **crucial importance for diagnosing and understanding** the mechanisms of autoimmune disorders. The clinical signs characteristic of autoimmune disorders are direct consequences of effector immunological processes. **The mechanisms** through which autoantibodies damage tissues and result in clinical signs are **many** (direct cytotoxicity, formation of immune complexes etc.)

Regardless of their pathophysiological mechanisms, autoantibodies are indisputably **sensitive and early markers of autoimmune disorders** and must therefore be used in any effort to make a diagnosis. They **can precede the appearance of clinical signs** by several months or even years (e.g.: anti-mitochondrial antibodies). **Some may be drug-induced** (e.g. antinuclear antibodies and

anti-TNF α biotherapy). Antibody titre varies as an autoimmune disease develops, but there is no necessary correlation between that variation and the clinical activity of the disorder. However, in some cases, repeat testing may be significantly useful in monitoring (e.g. anti-DNA antibodies and kidney damage due to lupus).

Autoantibodies are not necessarily pathogenic. They can be found in seemingly healthy individuals. Knowledge of the autoimmune origin of many conditions can help significantly improve treatment.

In practice

Diagnostic approach

In-depth clinical evaluation: Identify the evocative symptoms (persistent asthenia, arthralgia, skin irritation etc.)

Antibody testing: The most common tests relate to antinuclear, anti-DsDNA, anti-CCP and anti-thyroid antibodies. Guide your choice based on the clinical signs and suspected syndromes.

Cautious interpretation of results: The presence of autoantibodies is not necessarily a sign of a disorder. While screening for autoantibodies, supplementary identification tests may be added on the initiative of the pathologist. That individual is the main contact for interpreting autoantibody tests.

Performance of imagery examinations or biopsies: These explorations are often necessary for making a diagnosis or assessing tissue damage.

Multidisciplinary collaboration: If the results are positive or there is uncertainty, do not hesitate to refer the patient to a rheumatology or internal medicine specialist for an extended evaluation.

Key points

- Autoimmune disorders are the result of immune system dysfunction and can affect a wide range of organs and tissues.
- An autoantibody test is essential for diagnosing and managing autoimmune conditions.
- Collaboration between general practitioners, pathologists and specialists is crucial for providing optimum care to sufferers of autoimmune disorders.
- The detection and interpretation of autoantibodies calls for anin-depth understanding of their specificity and their clinical associations.
- For screening non-organ specific autoimmune disorders, prescribe ANAs and ANCAs.
- For screening organ specific autoimmune disorders, prescribe the **specific panel** of autoantibodies and indicate the **suspected syndrome**.



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Bibliography

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