Modern Methods of Laboratory Diagnosis of Autoimmune Diseases

Olimova Shaxnoza Shuxratovna

Clinical resident of the Bukhara Medical Institute. Uzbekistan

Abstract: Autoimmune diseases (AIDs) comprise a heterogeneous group of chronic disorders characterized by loss of immunological tolerance and the development of immune responses directed against self-antigens. Accurate and timely laboratory diagnostics play a pivotal role in early detection, differential diagnosis, prognostic assessment, and monitoring of therapeutic efficacy. Recent advances in laboratory immunology, including enzyme-linked immunosorbent assays, indirect immunofluorescence, immunoblotting, multiplex immunoassays, flow cytometry, and molecular genetic approaches, have significantly improved diagnostic accuracy and patient stratification.

Key words: autoimmune diseases, autoantibodies, laboratory diagnostics, ELISA, indirect immunofluorescence, immunoblotting, flow cytometry, molecular methods.

1. Introduction

TQM Autoimmune diseases represent a major challenge for modern healthcare systems due to their high prevalence, chronic and progressive course, and substantial impact on morbidity and quality of life [2,4]. Epidemiological data indicate that autoimmune disorders affect approximately 5–10% of the global population, with a steady increase in incidence observed over recent years. The clinical presentation of AIDs is often heterogeneous and nonspecific, particularly in early stages, making laboratory investigations essential for establishing a definitive diagnosis. Laboratory diagnostics of autoimmune diseases are primarily based on the detection of circulating autoantibodies, assessment of cellular immune dysregulation, and identification of genetic susceptibility markers [1]. Traditional diagnostic assays are increasingly complemented by advanced technologies that allow for improved sensitivity, specificity, and analytical throughput. Integration of these methods into diagnostic algorithms has significantly enhanced early diagnosis and stratification of patients [3].

Purpose of the Study. The aim of this review is to systematize contemporary laboratory diagnostic methods used in autoimmune diseases and to evaluate their clinical and diagnostic significance.

2. Materials and Methods

This review is based on an analysis of peer-reviewed publications indexed in international scientific databases, including PubMed, Scopus, and Web of Science. Original research articles, systematic reviews, meta-analyses, and international clinical guidelines published mainly within the last 10–15 years were included. A narrative and comparative analysis approach was applied to evaluate the diagnostic utility of different laboratory methods.

3. Results

Laboratory diagnostics of autoimmune diseases rely on a combination of serological, cellular, and molecular methods (table 1,2,3). Antinuclear antibodies (ANA) detected by indirect immunofluorescence on HEp-2 cells remain the primary screening tool for systemic autoimmune

rheumatic diseases. Anti-double-stranded DNA antibodies demonstrate high specificity for systemic lupus erythematosus and correlate with disease activity. Anti-cyclic citrullinated peptide antibodies are highly specific for rheumatoid arthritis and may be detected prior to clinical onset. Multiplex immunoassays allow simultaneous detection of multiple autoantibodies, while flow cytometry provides detailed immune cell profiling. Molecular genetic methods, including HLA genotyping, support risk stratification and personalized diagnostic approaches.

Table 1. Clinical significance of ANA fluorescence patterns

ANA pattern	Target antigens	Associated diseases
Homogeneous	dsDNA, histones	Systemic lupus erythematosus
Speckled	Sm, RNP, Ro/SSA	Sjogren's syndrome, MCTD
Nucleolar	RNA polymerase	Systemic sclerosis
Centromere	CENP-A, CENP-B	Limited systemic sclerosis

Table 2. Disease-specific autoantibodies

Disease	Autoantibodies	Diagnostic value
SLE	ANA, anti-dsDNA	High specificity
Rheumatoid arthritis	Anti-CCP	Early diagnosis
Sjogren's syndrome	Anti-Ro/SSA	Systemic involvement

Table 3. Laboratory methods in autoimmune diagnostics

Method	Application	Advantages	Limitations
IIF	ANA screening	High sensitivity	Subjective interpretation
ELISA	Autoantibody quantification	Automation	Cross-reactivity
Flow cytometry	Immune profiling	Functional analysis	Cost

4. Conclusions

Modern laboratory diagnostics of autoimmune diseases constitute a multi-level system integrating serological, cellular, and molecular methods. The combined application of these techniques enhances diagnostic accuracy, enables early disease detection, and supports individualized patient management.

Continued technological development, standardization of assays, and integration of novel biomarkers are expected to further improve diagnostic algorithms and clinical outcomes in autoimmune diseases.

References

1. Rose NR, Mackay IR. The Autoimmune Diseases. Elsevier; 2014.

2. Pisetsky DS. Antinuclear antibodies in rheumatic disease. Curr Rheumatol Rep. 2017;19:35.

3. Fritzler MJ. Multiplex diagnostic technologies in autoimmune diseases. Lupus. 2016;25:875–880.

4. Van Venrooij WJ et al. Anti-CCP antibodies. Nat Rev Rheumatol. 2011;7:391–398.

5. Tan WW, Li H, Chung I. Antibody Validation for Protein Expression on Tissue Sections. J Histochem Cytochem. 2020 May;68(5):325-333. doi: 10.1369/0022155420920866.

6. Weissferdt A, Kalhor N, Bishop JA, et al. Immunohistochemistry in the Diagnosis of Thoracic Tumors: A Practical Approach. Arch Pathol Lab Med. 2020 Sep 1;144(9):1015-1031. doi: 10.5858/arpa.2020-0072-RA.

7. Wolff AC, Hammond MEH, Allison KH, et al. Human Epidermal Growth Factor Receptor 2 Testing in Breast Cancer: American Society of Clinical Oncology/College of American Pathologists Clinical Practice Guideline Focused Update. Arch Pathol Lab Med. 2018 Nov;142(11):1364-1382. doi: 10.5858/arpa.2018-0902-SA.

8. Taube JM, Akturk G, Angelo M, et al. The Society for Immunotherapy of Cancer statement on best practices for multiplex immunohistochemistry (IHC) and immunofluorescence (IF) staining and validation. J Immunother Cancer. 2020 May;8(1):e000155. doi: 10.1136/jitc-2019-000155.

9. Topol EJ. High-performance medicine: the convergence of human and artificial intelligence. Nat Med. 2019;25(1):44–56.

10. Bossuyt PM, Reitsma JB, Bruns DE, et al. STARD 2015: an updated list of essential items for reporting diagnostic accuracy studies. BMJ. 2015;351:h5527.