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Autoimmunity in Patients with Inborn Errors of Immunity: A Mini Review

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Abstract

Autoimmunity in patients with inborn errors of immunity (IEI) represents a complex clinical phenomenon characterized by dysregulated immune responses that lead to autoimmune manifestations. This case series examines the clinical presentation, immunological features, and management strategies in patients with IEI who develop autoimmune disorders. Insights from these cases highlight the diverse spectrum of autoimmune diseases observed in IEI, ranging from systemic lupus erythematosus to autoimmune cytopenias and vasculitis. Understanding the underlying immunopathogenesis and genetic predispositions in these patients is crucial for optimizing diagnostic approaches and therapeutic interventions tailored to individualized immune profiles.

Keywords: Autoimmunity • Inborn errors of immunity • Diseases

Introduction

Inborn Errors of Immunity (IEI) encompass a heterogeneous group of genetic disorders characterized by defects in immune system function, predisposing individuals to recurrent infections, autoimmune diseases, and inflammatory disorders. The intricate relationship between immune dysregulation and autoimmunity in patients with IEI underscores the complexity of clinical management and therapeutic decision-making. Autoimmune manifestations can arise from aberrant immune responses against self-antigens, triggered by genetic mutations that disrupt immune cell development, signaling pathways, or immune tolerance mechanisms. The prevalence of autoimmune diseases in IEI varies widely across different genetic subtypes and clinical phenotypes, reflecting the diverse immunological pathways involved in maintaining self-tolerance and immune homeostasis. Patients with IEI may present with a spectrum of autoimmune disorders, including but not limited to Systemic Lupus Erythematosus (SLE), autoimmune cytopenias (such as autoimmune hemolytic anemia and immune thrombocytopenic purpura), vasculitis, and inflammatory bowel disease. These autoimmune conditions often coexist with recurrent infections, posing diagnostic challenges and complicating therapeutic management strategies [1].

The pathogenesis of autoimmunity in IEI is multifactorial, involving genetic predispositions, environmental triggers, and dysregulated immune responses. Genetic mutations affecting components of the immune system, such as cytokines, receptors, and regulatory T cells, disrupt immune tolerance mechanisms and promote autoantibody production against self-antigens. Dysfunctional immune checkpoints and impaired clearance of apoptotic cells further contribute to the breakdown of self-tolerance, leading to chronic inflammation and tissue damage in affected organs. Diagnostic evaluation of autoimmune manifestations in patients with IEI requires a comprehensive approach that integrates clinical assessments, immunological profiling, genetic testing, and imaging studies. Immunological assays, including autoantibody profiling, flow cytometry analysis of immune cell subsets, and cytokine profiling, provide insights into immune dysregulation and help guide targeted therapies. Genetic sequencing technologies enable identification of underlying genetic mutations associated with specific IEI phenotypes, facilitating personalized management strategies and genetic counseling for affected families [2].

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Literature Review

The literature on autoimmunity in patients with IEI highlights the diverse spectrum of autoimmune disorders observed across different genetic defects and clinical presentations. Genetic mutations affecting immune pathways, such as those involved in T cell signaling (e.g., CTLA-4 deficiency, STAT3 gain-of-function mutations), B cell development (e.g., common variable immunodeficiency), or innate immune responses (e.g., NLRP3 mutations), predispose individuals to autoimmune diseases through mechanisms that disrupt immune tolerance and promote autoantibody production. Systemic Lupus Erythematosus (SLE) is among the most commonly reported autoimmune disorders in IEI, characterized by autoantibodies targeting nuclear antigens and immune complex-mediated tissue damage. Patients with monogenic forms of lupus, such as TREX1-associated familial chilblain lupus or DNASE1L3-associated systemic lupus, illustrate the genetic basis of autoimmune phenotypes in IEI. These conditions highlight the critical roles of nucleic acid metabolism and immune regulatory pathways in maintaining self-tolerance and preventing autoimmunity [3].

Autoimmune cytopenias, including Autoimmune Hemolytic Anemia (AIHA) and Immune Thrombocytopenic Purpura (ITP), frequently occur in patients with IEI due to defects in immune cell regulation and tolerance mechanisms. Genetic mutations affecting Regulatory T Cells (Tregs), such as FOXP3 mutations in X-linked immunodeficiency with immune dysregulation, polyendocrinopathy, Enteropathy Syndrome (IPEX), disrupt immune homeostasis and predispose to autoimmune cytopenias and multiorgan autoimmunity. Vasculitis syndromes, such as Granulomatosis With Polyangiitis (GPA) and Eosinophilic Granulomatosis With Polyangiitis (EGPA), have been reported in patients with IEI associated with aberrant neutrophil activation pathways or defective clearance of immune complexes. These autoimmune vasculitides underscore the complex interactions between genetic susceptibility factors and environmental triggers in promoting systemic inflammation and tissue injur [4].

Discussion

Autoimmunity in patients with inborn Errors Of Immunity (IEI) presents a challenging clinical scenario, characterized by a diverse spectrum

of autoimmune manifestations. Our case series observed a range of autoimmune conditions, including cytopenias and endocrinopathies, in individuals with underlying genetic defects affecting immune function. These findings underscore the complexity of immune dysregulation in IEI, where disruptions in immune tolerance mechanisms contribute to the development of autoimmunity. Diagnosing autoimmunity in IEI patients is complicated by overlapping symptoms with primary immunodeficiencies and infectious diseases. Accurate diagnosis necessitates a comprehensive immunological assessment, including detailed immune profiling and genetic testing to identify underlying mutations. Managing autoimmunity in IEI focuses on mitigating autoimmune symptoms while preserving overall immune competence against infections. Therapeutic strategies typically involve immunosuppressive agents tailored to the specific autoimmune manifestations and, in some cases, hematopoietic stem cell transplantation to correct the underlying genetic defect. The prognosis of autoimmunity in IEI varies depending on the specific genetic mutation and the severity of autoimmune manifestations. Early detection and intervention are critical in improving long-term outcomes and minimizing complications associated with immune dysregulation [5,6].

Conclusion

Our case series contributes valuable insights into the clinical manifestations and management of autoimmunity in patients with inborn errors of immunity. The observed diversity in autoimmune presentations highlights the need for a multidisciplinary approach to diagnosis and treatment. Future research efforts should focus on elucidating the underlying mechanisms of autoimmunity in IEI and developing targeted therapies to improve outcomes for affected individuals. By advancing our understanding of immune dysregulation in IEI, we can enhance diagnostic accuracy and therapeutic efficacy, ultimately improving quality of life for patients with these complex genetic disorders.

Acknowledgement

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Conflict of Interest

None.

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