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Primary Immunodeficiencies: Pathogenetic Advances, Diagnostic and Management Challenges

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Message from the Guest Editors

In the last decade, the research advances in the field of pediatric immunology brought a dramatic improvement in the diagnosis of primary immunodeficiency disorders. The use of new genetic panels allowed the identification of a considerable number of monogenic causes of primay immunodeficiency, each one featuring a peculiar clinical and immunological phenotype.

Moreover, the discovery and molecular characterization of the monogenic causes of primary immunodeficiencies helped to better understand the complex pathogenic and clinical overlap between immunodeficiency, immune dysregulation, autoimmunity and autoinflammation. The risk of lynphoproliferation and non-hematologic malignancies in children with primary immunodeficiencies has also been deeply analyzed, providing interesting information for long-term follow-up and clinical management.

The present Special Issue aims to provide an update on the intriguing field of primary immunodeficiency disorders in childhood, with a particular focus on the clinical, immunological and molecular diagnostic aspects and on the most relevant improvements in the therapeutic approach to such conditions.













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