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Primary Immunodeficiencies: Pathogenetic Advances, Diagnostic and Management Challenges

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This Special issue focuses of the most relevant advances in the field of inborn errors of immunity. Indeed, as immunology is rapidly progressing, with the constant discovery of new disease entities and the better characterization of molecular mechanisms and targeted therapies, a continuous update is mandatory for clinician and researchers. In this Issue, the non-infectious manifestations of inborn errors of immunity (such as autoimmunity, atopy, and lymphoproliferation) are deeply discussed, as well as the impact of the novel genetic sequencing techniques in the diagnostic approach. Moreover, some well-known disorders, including common variable immunodeficiency, are extensively reviewed and revisited in a modern way. Also, diseases of recent description, including some disorders of immune dysregulation and inborn errors of immunity associated with syndromes, are described. Finally, some of the papers of this Special Issue focus in a detailed way the molecular mechanisms leading to immunodeficiency and immune dysregulation, also opening the way for further clinical and preclinical studies.

As the expanding availability of immunological and genetic testing offers the opportunity to identify new disease entities and elucidate the function of new genes involved in the development and regulation of the immune response, we aim to present the most relevant innovations in this delicate field.



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