IMMUNOLOGY

DISORDERS OF IMMUNE REGULATION

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Abstract: *Inborn errors of immunity are genetic disorders with broad clinical manifestations, ranging from increased susceptibility to infections to significant immune dysregulation. As per 2019 Update of the International Union of Immunological Societies expert committee's classification, there are now 430 single-gene inborn errors of immunity. Primary immune regulatory disorders are a growing subset of diseases referred to as inborn errors of immunity. Unlike classical primary immune deficiency disorders that typically present with severe, recurrent, or unusual infections, the clinical manifestations of primary immune regulatory disorders are dominated by immune-mediated diseases (autoimmunity, autoinflammation/ hyperinflammation, lymphoproliferation, malignancy, and severe atopy). In this article we will discuss in detail about disorders of immune regulation with phenotypical presentation and associated genetic defects.*

Keywords: *Immune deficiency, Inborn errors of immunity, Primary immune regulatory disorders, Autoimmunity.*

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Points to Remember

- ***PIRD predominantly have clinical features of autoimmunity, hyperinflammation, lympho-proliferation, malignancy and severe atopy with less dominant features of immunodeficiency and infection.***
- ***Genetic causes of PIRD function in immune pathways that regulate the various types of immune responses.***
- ***The treatment is challenging, as it requires careful balancing of immunosuppression in subjects at increased risk of infections.***
- ***Treatment for PIRD are directed at the specific genetic defect, and HCT can be a curative therapy for some cases***

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