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INBORN ERRORS OF IMMUNITY IN CLINICAL PRACTICE - A PATTERN BASED PRACTICAL APPROACH

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Abstract: *Inborn errors of immunity, formerly known as primary immunodeficiency disorders, comprise over 550 genetically defined conditions characterized by immune dysregulation including autoimmunity leading to susceptibility to infection, autoinflammation, lymphoproliferation, and malignancy. Early recognition relies primarily on clinical suspicion and pattern recognition rather than advanced molecular diagnostics. Severe, persistent, unusual, or recurrent infections, early-onset autoimmunity, unexplained cytopenias, chronic inflammation, and vaccine-related complications should prompt evaluation. Initial assessment with complete blood count, differential counts, and peripheral smear often provides key clues guiding immunoglobulin assays, lymphocyte subset analysis, neutrophil oxidative burst testing, complement evaluation, and genetic studies. A phenotype-driven, cell-based diagnostic approach facilitates early referral.*

Keywords: *Inborn errors of immunity, Primary immunodeficiency, Pattern recognition, Recurrent infections, Autoimmunity, Pediatric immunology.*

Points to Remember

- ***Inborn errors of immunity (IEI) are not uncommon among children admitted with severe, recurrent, unusual or persistent infections.***
- ***Non-infectious clues like autoimmunity, immune dysregulation and inflammatory patterns are increasingly recognized as IEI presentations.***
- ***Pattern recognition remains the cornerstone of early diagnosis.***
- ***Secondary causes, particularly HIV infection, must be excluded before labelling IEI.***
- ***A simple complete blood count (CBC) with differential often provides crucial clues to the immune cell involved and helps guide targeted immunological testing.***
- ***Early diagnosis alters outcomes - curative options such as hematopoietic stem cell transplantation and disease-modifying therapies including IVIG replacement and biologics are increasingly available.***
- ***In IEI, vaccine decisions must be individualized***

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