

## IMMUNOLOGY

### LABORATORY CLUES TO PRIMARY IMMUNODEFICIENCY DISORDERS

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**Abstract:** Primary immunodeficiency disorders are a large group of heterogeneous diseases, which result from defects in the immune system. These defects can either be in the innate or adaptive immunity. As per the latest classification published by the International Union of Immunological Societies expert committee on Inborn Errors of Immunity, around 430 primary immunodeficiency disorders have been recognized and the list is expanding. One in 1000 individuals suffer from a primary immunodeficiency disorder and these diseases are by no means, rare. Yet many patients remain undiagnosed, due to lack of awareness of these conditions. This article highlights the importance of routine tests like complete blood counts and serum immunoglobulin assay in diagnosing patients with these disorders. Along with case-based discussion, simple algorithms have been provided, that can guide a clinician in making a timely diagnosis.

**Keywords:** Primary immune deficiency diseases, Inborn errors of immunity, Laboratory tests.

### Points to Remember

- *PIDs are currently referred to as IEs.*
- *One in 1000 individuals suffer from a PID, hence, these diseases are not rare.*
- *The first step in the diagnosis of PIDs is history and clinical examination and to suspect them in clinical practice.*
- *Careful analysis of routinely available tests like hemogram provide valuable clues to the underlying PID.*
- *Neutropenia can be manifestation of a PID and noted in severe congenital neutropenia, cyclic neutropenia, hyper IgM syndrome and many other PIDs.*
- *Marked neutrophilia must make one think of leukocyte adhesion deficiency.*
- *Persistent thrombocytopenia in a male child warrants evaluation for possible Wiskott Aldrich Syndrome.*
- *Serum immunoglobulin assay is a valuable tool in evaluation of patients with suspected PID, which has to be used more often in clinical practice.*
- *NBT and DHR are simple screening tests for chronic granulomatous disease.*
- *Once suspected, one must not delay initiation of therapy for want of genetic testing in these patients.*

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