IMMUNOLOGY

UTILITY OF GENETIC TESTS IN PRIMARY IMMUNODEFICIENCY DISORDERS

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Abstract: *Genetic testing plays a crucial role in the field of primary immunodeficiency. It provides the confirmatory molecular diagnosis to the affected patient. This helps the family in prenatal diagnosis, personalized treatment, embryo implantation during in-vitro fertilization and family screening. In this review, we have broadly discussed the widely used genetic tests in the clinical setting for primary immunodeficiency. We have also described the most appropriate genetic testing approach for different types of primary immunodeficiency. the utility of genetic testing to the affected patients and their family members is also discussed.*

Keywords: *Primary immunodeficiency, Genetic testing, Molecular diagnosis, Personalized treatment.*

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Points to Remember

- ***Genetic testing provides the confirmatory diagnosis for the patients affected with primary immunodeficiency that has a heterogeneous array of symptoms.***
- ***The identification of the variant helps the clinicians in tailoring the treatment of the patient according to the genetic condition.***
- ***Variant identification helps in prenatal diagnosis, embryo pre implantation, family and community screening.***
- ***Choosing the most appropriate genetic test for diagnosis of different types of PID is based on the patient's clinical characteristics and immunological investigations.***

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