

# **Primary immune deficiency disorders in the South Pacific: the clinical utility of a customized genetic testing program in New Zealand**

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### **ABSTRACT**

Primary immune deficiency disorders (PIDs) are a group of diseases associated with a genetic susceptibility to recurrent infections, malignancy, autoimmunity, and allergy. The molecular basis of many of these disorders has been identified in the last two decades. Most are inherited as single gene defects. As discussed in this paper, identifying the underlying genetic defect plays a critical role in many areas—including patient management, diagnosis, identifying atypical presentations, family studies, providing prognostic information, prenatal diagnosis, and defining new diseases. New Zealand is a geographically isolated, developed country in the South Pacific. We have introduced a dedicated customized genetic testing service for PID patients in New Zealand. This accredited diagnostic program offers rapid turnaround times for genetic tests and minimizes the risk of laboratory errors. Here we review the clinical indications for genetic testing for PIDs based on cases referred to the molecular immunology diagnostic service at Auckland City Hospital.

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