الاختبارات الجينية في ض.م.أ: لا بد منها ؟

Genetic testing in PID: Is it a must

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Primary immunodeficiency disorders (PIDs) comprise more than 200 different disorders that affect the development, function, or both of the immune system. In most cases, PIDs are monogenic disorders that follow a simple Mendelian inheritance; however, some PIDs are of more complex polygenic origin. Higherincidence rate is observed among populations with high consanguinity or among genetically isolated populations.The application of mutation analysis is becoming an integral part of the complete evaluation of patients with primary immunodeficiencies. Mutation analysis can (1) provide a definitive diagnosis; (2) help in genetic counseling, permitting early prenatal diagnosis and carrier identification; (3) establish a diagnosis in atypical presentations; (4) provide relevant information in selected PIDs in which there is a strong genotype-phenotype correlation so that prognostic implications can be derived; and (5) permit pre-symptomatic identification of individuals affected with potentially lethal forms of PIDs and hence prompt timely life-saving interventions, such as hematopoietic cell transplantation (HCT).Genomic DNA sequencing is currently the standard approach used to characterize a possible gene mutation causing a specific primary immunodeficiency. Here we discuss the importance of and the limitations associated with molecular diagnosis of these disorders and emphasize the need that mutation analysis be accompanied by appropriate evidence that the identified genetic defect has pathologic consequences.

زواج الاقارب و نقص المناعة الاولية

Consanguinity and Primary Immunodeficiencies

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Primary immunodeficiencies (PIDs) are a heterogeneous group of genetic disorders caused by defects in the immune system that predispose patients to infections, autoimmune disease, lymphoproliferation and malignancy. Most PIDs are inherited in an autosomal recessive pattern, and they are therefore more common in areas with high rates of consanguineous marriage. Reports about PIDs from these areas have demonstrated a peculiar prevalence of more severe forms of disease compared to other regions, and patients born to consanguineous parents have increased rates of morbidity and mortality compared to other patients. Individuals at high risk of having a child with a PID who wish to have a healthy child have limited options, which include prenatal diagnosis and pre-implantation genetic diagnosis. However, these options require a collaborative team of specialists and may not always be implemented due to geographic, religious, financial or social factors. There is a need for the implementation of strategies to increase public awareness of the health risks associated with consanguineous marriage. It should be stressed that genetic counseling should be an important component of the care of patients with PIDs as well as their families.