Human DNA: our genetic encyclopaedia

The human body is made up of many different types of cells, most of which contain chromosomes. The chromosome is composed of tightly coiled DNA strands. DNA contains essentially all the information to make a human being. The information is 'stored' in genes. When the genes are activated, proteins can be made and transported to where they are needed.

The human DNA codes for 31,000 to 39,000 genes. Each gene is present in two copies, one from the father and one from the mother. The combined expression of all the genes determines everything that makes each one of us unique (height, eye colour, hair colour and so on).

What is genetic testing?
Genetic testing involves examining a person's genes, chromosomes or proteins to detect a heritable disorder or disease. A test sample is usually blood, but sometimes can be tissue or body fluid.

Genetic testing at LabPlus, Auckland City Hospital
The molecular immunology section at LabPlus offers primary immunodeficiency genetic testing services through healthcare providers such as doctors and genetic counsellors. However, we do not offer direct-to-consumer genetic testing.

Genetic testing of primary immunodeficiency disorders
Some primary immunodeficiency disorders are due to DNA changes of a single gene. However, some disorders such as Common Variable Immunodeficiency could be due to defects in a number of genes.

Primary Immunodeficiency Disorders	Defective gene
X-linked lymphoproliferative syndrome (XLP)	SH2D1A
X-linked Agammaglobulinaemia (XLA)	BTK
Common Variable Immunodeficiency (CVID)	CD19, BAFFR, TACI, ICOS

What is involved in genetic testing?
DNA is extracted from patient's blood and stored at -20 °C until use. Since it is difficult to work with two copies of a gene, we employ a technique known as Polymerase Chain Reaction (PCR) to help us out! PCR is like a photocopying machine; it allows us to make many copies of the gene we want to examine. Once a sizable pool of gene copies is achieved, we can then determine the DNA sequences of the gene with a special instrument called ABI 3100 Genetic Analyser. DNA sequences of the patients will be compared to healthy individuals using a computer alignment programme. This process will assist with identifying mutations. The laboratory reports the test results in writing to the patient's doctor or genetic counsellor.

PCR machine

ABI 3100 Genetic Analyser

Not all DNA mutations are harmful
It is important to remember that not all DNA mutations are harmful. Every human carries variations of DNA sequence in their genes, which is known as polymorphism. Only mutations that disrupt the function of the gene product (or protein) are potentially harmful.

Benefits of genetic testing
Identification of a gene defect can assist with patient management. Genetic testing can also assess family members at risk of inheriting a disorder. Carrier testing could help couples determine if they have the defective gene that could be passed on to the next generation. Other benefits include pre-natal diagnosis if required and a better understanding of the immune system.

Limitations of genetic testing
Genetic testing has its limitations. It can only detect small DNA changes, deletions or insertions within the gene. Gross chromosome deletions or DNA changes outside the gene of interest that may influence the outcome of disorders cannot be easily detected.

Informed Consent
Patients usually undergo counselling before being offered genetic testing, so that they are able to make an informed decision about testing. It is important that the patients understand the testing procedure, the benefits and limitations of the test, and the possible consequences of the test results.

Who is eligible for genetic testing?
All individuals and families are eligible for testing after consultation with their doctor and the genetic counsellor. It is not possible to do specific tests for all disorders so testing is subject to availability.

Glossary

DNA: the total genetic material that define the making of an organism

Chromosomes: All the genetic material are organised into 23 pairs of chromosomes. An analogy would be all written material that defines English language are organised into 23 volumes of encyclopaedia.

Gene: a segment of DNA that codes for a component important for an organism to function

Protein: the component that carries out important function in an organism. Different proteins carry out different functions in the body.

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These booklets are designed to offer medical professionals, patients and their families' basic information about these rare disorders of the immune system.
For further information please contact your immunologist, paediatrician, physician or the National IDF Health Coordinator

Other booklets available:

Living with PID
What is IVIG Therapy
Recurrent infections
Common Variable Immune deficiency (CVID)
X-linked Agammaglobulinaemia (XLA)
Chronic Granulomatous disorder (CGD)
Selective IgA Deficiency

IDF
Immune Deficiencies Foundation
Asia Pacific Alliance

The Immune Deficiency Foundation Asia-Pacific Alliance, IDFAPA.

An alliance of not-for -profit PID Patient support groups across the Asia Pacific Region.

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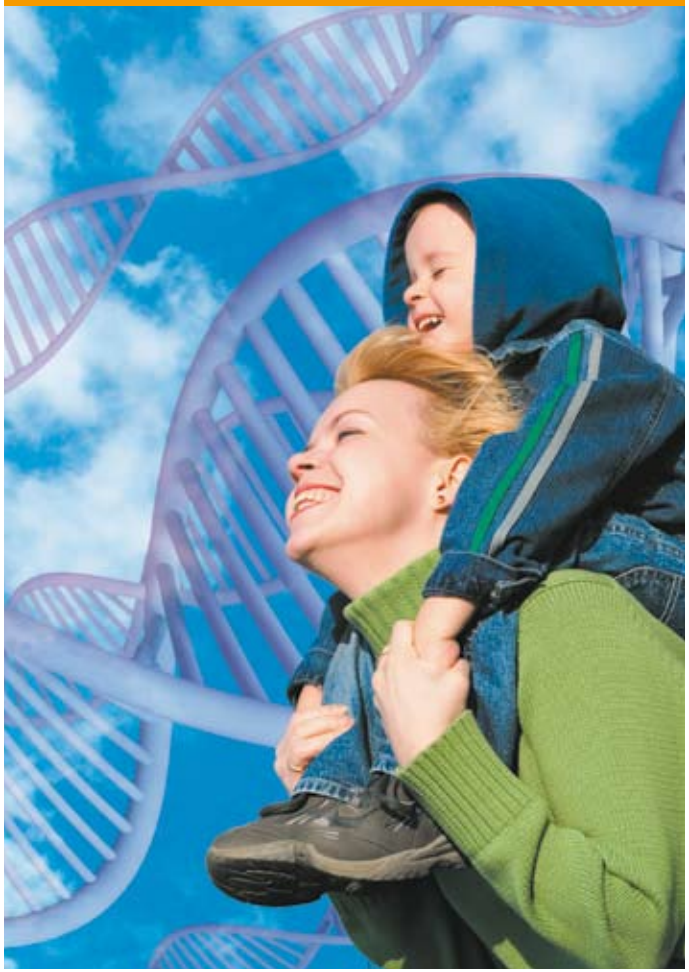
Living with Primary Immune Deficiency Disorders

Genetic Testing and PID



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