



AUSTRALIA AND NEW ZEALAND
ANTIBODY DEFICIENCY ALLELE STUDY

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Background

Significant progress has been made in the genetics of Primary Immunodeficiency Disorders, including antibody deficiency disorders. Nevertheless, in the majority of cases of primary antibody deficiency, the genetic basis remains obscure and pathogenesis is not understood. A small proportion (<10%) of patients with Common Variable Immunodeficiency (CVID) or lesser forms of antibody deficiency such as Specific Antibody Deficiency have identified gene defects.

The Australia and New Zealand Antibody Deficiency Allele (ANZADA) will build on the achievements of the ASCIA Australia and New Zealand PID Register, to investigate the genetic basis of primary antibody deficiency.

Aims of the study

The ANZ-ADA study has four primary aims:

1. To determine the prevalence of known CVID-associated gene defects in Australia and New Zealand.
2. To discover novel gene defects through screening patients for alleles identified in mouse models of antibody deficiency.
3. To define cellular phenotypes correlated with identified CVID-associated alleles.
4. To establish the Australia and New Zealand DNA repository for patients with PID.

Methods

This study will utilise the ASCIA PID Register to identify patients with antibody deficiency disorders who will be invited to participate. DNA samples will be collected, and clinical data captured on the register will be linked with the DNA repository. DNA will be screened for mutations and polymorphisms in known disease-causing alleles and those with no known mutations identified will be analysed for novel genetic mutations in candidate genes. Screening will be performed using both PCR and high resolution melting assays.

Dr Matthew Cook (Principal Investigator) and Dr Carola Vinuesa (Associate Investigator) already have extensive experience in investigation and discovery of novel genetic mutations in human immune disease, and this project will adopt similar gene identification processes as those already established by these two investigators.

Research collaborations

A network of Immunologists from across Australia and New Zealand will be involved in patient recruitment. During the initial stages of the project, samples will be collected from The Canberra Hospital, Concord Hospital, Westmead Hospital, Royal Prince Alfred Hospital, Royal Melbourne Hospital, John Hunter Hospital and The Childrens' Hospital, Westmead. Expansion into other states and New Zealand is anticipated, with the timing of this dependant on project funding. Investigators have already established collaborations with international research groups and Immunologists and opportunities for international collaborations in this project will be investigated in the future.

The establishment of a DNA repository will be a major advance in PID research in Australia and New Zealand. It is anticipated that other research groups will make applications for access to the specimens held in this repository, increasing awareness and interest of this project to a national and international scale.

Further information

If you would like further information about this study, please go to the website (www.anzada.org) or contact the study coordinator, Philippa Kirkpatrick (details above).